Chapter 1

Genetic Counseling and the Physician–Patient Relationship

OVERVIEW

Medicine’s ethical maxim, primum non nocere—above all do no harm—and its underlying values and objectives of treatment and cure, traditionally have been regarded as the fundamental role and value of medicine in society [Gadow, 1984; Jonsen et al., 1998]. Within the context of genetic testing, the goals and values adopted in the practice of counseling need to reflect a variety of applications and settings. The lack of effective treatment and cure for many genetic diseases and disorders has triggered significant debate concerning what objectives and values constitute an appropriate model of care. Accordingly, it is appropriate to consider how the traditional medical concepts of “benefit,” “harm,” and the physician–patient relationship have been interpreted within the context of genetic testing and to look at practical steps that can be taken to ensure that the patient benefits from counseling.

INTRODUCTION

Genetics has moved beyond the scope of individual families and quiet conversations about which uncle might have had what problem. With widespread uptake of prenatal screening, expansion of newborn screening programs, the advent of conclusive laboratory diagnostics for many developmental disorders of childhood, predictive testing and risk assessment for disorders of adult onset, new reproductive technologies, cancer diagnostics, the widely promoted promises of biotechnology, and the immediate accessibility of Internet-based information, patients have moved from passive to active users of genetic services. As genetic components of common diseases become...
recognizable, quantifiable, and predictable, risk modification techniques will be extended to whole populations. We are all now the patient. Governments, professionals, private enterprise, and the lay press have responded to this situation, and the result is more information (accurate or not) from more sources (biased or not) available to more patients (receptive or not) every day.

To return to the clinic, this movement of genetics into the mainstream of health care and public debate has changed the expectations of our patients. However, for many situations we still face an unfortunate technological lag between our theoretical knowledge of a given genetic disease and our systemic ability to provide effective therapy. Thus, we not only frequently have a knowledgeable patient but often also a frustrated one. Genetic counseling is by definition an interactive process. More than ever, health care professionals will have to assess how much the patient can comprehend about the situation at hand and be skilled at both extracting and providing critical information that will benefit the patient in decisions about care. Clearly, communication skills and the fundamental nature of the relationship between the patient and the provider are very important.

Why has the patient sought professional services? While concern and worry about a perceived medical problem are a common motivation to seek medical attention, the specific components of a patient’s expectations might become particularly important if there is no effective treatment for the disease in question. Discussion can clarify what the patient expects to learn, uncover the patient’s psychosocial status and cultural and family context, correct misconceptions, assess concerns, and provide an opportunity to discuss the nature, objectives, risks, and limitations of genetic testing. The patient needs a realistic understanding of the implications posed by a positive, negative, or inconclusive result. A failure to review these issues may contribute to patient misunderstanding and dissatisfaction, which are both critical factors in a patient’s decision to seek redress through malpractice litigation.

**CORE CONCEPTS**

The patient–health care professional relationship in genetic counseling is:

- Interactive
- Respectful of patient autonomy (nondirectiveness and incorporation of patient values)
- Client centered
- Founded on the goals of achieving supportive patient education, enabling an informed decision, providing accurate risk assessment and risk perception, facilitating decisions, attending to psychosocial needs for the patient and the family, empowering the patient, and facilitating future care

Health care professionals need to adapt their counseling techniques and goals to a variety of applications and indications, including type of practice (clinical setting) and
type of indication [diagnostic confirmation, at-risk testing (carrier status), prenatal screening, population screening, presymptomatic testing, susceptibility testing].

Patient motivation to participate in and act upon genetic counseling is influenced by the patient’s perception of risk and effectiveness of clinical management (treatment). Patients accrue knowledge from many different sources. Patients may access information that is of dubious origin, authenticity, or benefit. Patients may not be able to critically appraise the clinical relevance of the information they obtain and may either seek guidance about their information or use it as an opinion in contrast to the genetic counseling they receive. The patient’s beliefs, knowledge, and cultural values may conflict with the goals and expectations of a nondirective counseling process. Health care professionals may need to prepare the patient for counseling about test results.

Genetic counseling is often provided at a time of emotional stress. The timing and setting for counseling may prevent the patient from understanding what is said. The process of risk assessment, understanding risk, and modifying perception of risk may be difficult for patients to utilize (see also Chapter 14, Test Results: Communication and Counseling). The psychological status of the patient needs to be assessed so that effective communication can ensue (see also Chapter 2, Communication).

Health care practitioners need to understand the limitations and applications of genetic testing and predictions of clinical significance and be able to accurately convey that information to the patient (see also Chapter 7, Informed Consent). However, health care professionals who have not received specialized training in genetics may be uncertain as to the nature and extent of their responsibilities [Veach, 2004].

Patients may make management decisions based upon their own frame of reference. The patient should expect the freedom of informed choice in regard to management decisions, and the health care provider has to accept that some patients will make choices that do not appear to be based upon the information provided.

**TRUST, GENETIC TESTING, AND THE PHYSICIAN–PATIENT RELATIONSHIP**

When a patient is faced with a medical illness or problem, the patient asks the physician to find an appropriate solution, usually in terms of treatment and cure. In seeking the physician’s aid, the patient must provide the physician with a certain degree of trust. The patient must have confidence that the physician will be able to resolve the medical difficulty, will exercise good judgment, and will utilize medical skills and knowledge with due care, diligence, and caution [Sharpe, 1997; Mechanic, 1998]. In times of emergency, uncertainty, and anxiety, the patient may be especially dependent upon the physician to come to her or his aid. Due to their medical knowledge and skills, physicians perceive themselves as trustworthy and assume that because their patients trust and have confidence in them, a reasonable patient will, or should, adopt the recommended course of action because the physician has determined that
Chapter 1 Genetic Counseling and the Physician–Patient Relationship

it provides the best opportunity for therapeutic benefit and the prevention of medical harm [DuBose, 1995; Sharpe, 1997].

“Trust,” within the context of this type of physician–patient relationship, reflects medicine’s ethical maxim, primum non nocere—above all do no harm—and its underlying values of treatment and cure. Given these perceptions and values, the physician–patient relationship traditionally was perceived as one that reflected an inherent disparity in medical knowledge and expertise. Accordingly, the nature of the relationship was deemed to be paternalistic, the patient dependent and passive, the physician paternal, active [Katz, 1984; Apfel and Fisher, 1984; Gillon, 1986], and best able to determine “what constitutes well-being” [Katz, 1977, 1993; Infelfinger, 1980; Kinsella, 1988].

The traditional therapeutic model of care, first proposed by René Descartes in the seventeenth century, was based on the idea that the human body was a machine and functioned pursuant to discernible mechanical principles. Descartes separated the soul—the patient’s beliefs, needs, fears, and values—from the bodily processes and argued that such emotional concerns had little in common with physical health and, therefore, the care and cure of bodily problems [Carter, 1983]. Late into the twentieth century, medical and social commentators argued that the Cartesian biomedical model remained the dominant therapeutic model, exemplifying medicine’s traditional values of “the relief of pain, the prevention of disability, and the postponement of death by the application of theoretical knowledge incorporated in medical science” [Seldin, 1981; Krauser, 1989]. This included the operative assumption that disease can be fully accounted for by “deviations in measurable biological variables,” without reference to the “social, psychological, and other behavioral dimensions of illness” [Engel, 1991].

However, many of these operative assumptions, and their underlying values, increasingly were questioned and challenged [Lynch, 1985; Englehardt, 1986; Gillon, 1986]. In the last quarter of the twentieth century, it was argued that the physician–patient relationship had become less paternalistic with increased shared decision making, as patients took on a great responsibility and demanded more autonomy in medical decision making [Balint and Shelton, 1996; Charles et al., 1997, 1999; but see Zupancic et al., 2002]. In 2003, a World Health Association survey of 3707 patients and doctors in the United States, United Kingdom, Canada, Germany, South Africa, and Japan reported that only about 20 percent of those surveyed defined the physician–patient relationship as paternalistic or authoritarian [Pincock, 2003]. Health Maintenance Organizations (HMOs) and the advent of “managed care,” however, continue to have a dramatic impact in the United States. One study reported that nearly one in three doctors in the United States withheld information from patients about medical services because they were not covered by health insurance plans [Wynia et al., 2003; Gottlieb, 2003; Mechanic, 1998].

Many types of genetic tests may not clearly promote diagnosis, treatment, and cure. They may only provide information about a medical condition that is likely to occur at some time in the future. Testing may fail to predict how severe the medical condition may be, when it will occur, or even, due to reduced penetrance, that it
Genetic Counseling and the Physician–Patient Relationship

will occur [Andrews and Zuiker, 2003]. These limitations can give rise to complex and troubling ethical and moral issues. By way of example, should a health care professional recommend adoption or reproductive technology in order to prevent the transmission of a genetic disorder [Shaw, 1987]? If so, what type of genetic disease or disorder justifies a recommendation of adoption, or reproductive technology, or termination of a pregnancy; what is the appropriate recommendation, if any, in the case of hereditary deafness [Taneja et al., 2004] or achondroplasia [Green, 1997] or mental conditions [Andre et al., 2000; compare to Vehmas, 2002] (see also section by Mykitiuk et al. in Chapter 8, Prenatal and Neonatal Screening)?

Because of these limitations, physicians can be confounded by patient anguish, anxiety, and despair. In these circumstances, the patient may feel helpless and alone, and the physician also may experience a sense of powerlessness. In such a context, the *traditional* perception of medical trust is reduced to an illusion [DuBose, 1995].

If health care professionals are to provide benefit and prevent harm to the patient before, during, and after genetic testing, they will need to change the way they perceive the nature of their relationship with the patient and how they can best respond to the patient’s pleas for assistance and relief [Maley and NGSC, 1994; Sharpe, 1997]. Health care professionals need to focus on a patient’s communicative, emotional, and psychological needs in order to assist the patient to understand, adjust to, and cope with the implications posed by genetic testing and test information [Ad Hoc Committee on Genetic Testing, 1975; Sandhaus et al., 2001]. In short, health care professionals have to apply a therapeutic model of care that encompasses a more “human vision” as opposed to a purely Cartesian “medical vision” [DuBose, 1995; and see also section by Mykitiuk et al. in Chapter 8, Prenatal and Neonatal Screening].

**GENETIC COUNSELING AND THE PHYSICIAN–PATIENT RELATIONSHIP**

Genetic counseling represents a significant departure from other forms of medical care in not giving overt advice concerning decision making by a patient. Given the lack of effective treatment and cure for many genetic diseases and disorders, the pivotal factor in decision making can be the nature of the genetic test information and its impact on the patient and the family, including—as will be discussed in later chapters—the patient’s objectives, concerns, values, and beliefs, as well as the potential for insurance and employment discrimination, stigmatization, and the psychological implications [Wiggins et al., 1992; Tibben et al., 1993a; Michie et al., 1997a; Michie and Marteau, 1999b; Codori et al., 1999; Glanz et al., 1999; Kirschner et al., 2000; Bassett et al., 2001; Hadley et al., 2003; Keller et al., 2004].

Whether seen from the perspective of good patient care and/or reducing the risk of patient misunderstanding, dissatisfaction, and allegations of malpractice, genetic counseling and the specialized practice of genetic counselors, should be considered
a critical, if not mandatory, component of the testing process. Counseling:

- Provides improvement in risk estimation and comprehension [Lerman et al., 1997; Burke et al., 2000; Edwards et al., 2000].
- Promotes discussion of risk management options [Johnson et al., 2002].
- Facilitates quality assurance [Gibons, 2004], better understanding, more informed decision making, and helps to reduce exposure to malpractice allegations.

Effective counseling helps the patient and family to: (1) comprehend the medical facts including the diagnosis, probable course of the disorder, and the available management, (2) appreciate the way heredity contributes to the disorder and the risk of recurrence in specified relatives, (3) understand the alternatives for dealing with the risk of recurrence, (4) understand, cope with, and adapt to the emotional and psychological aspects of testing and test results; (5) choose a course of action that seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards; and (6) make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder [Ad Hoc Committee on Genetic Counseling, 1975; Royal Commission, 1993; Burgess et al., 1998].

NONDIRECTIVENESS

A comprehensive review of practitioner surveys and human/medical genetics journal and text literature indicates that a significant majority of geneticists and counselors subscribe to the ethical value of nondirective counseling [Ad Hoc Committee on Genetic Counseling, 1975; Kessler, 1979; Fletcher et al., 1985; Wertz and Fletcher, 1988; Wertz et al., 1990; De Dinechin et al., 1993; Committee on Assessing Genetic Risk, 1994; Benkendorf et al., 1997; Biesecker, 2001; Kessler, 2001; but see Yarborough et al., 1989; Clarke, 1991; Royal Commission, 1993; Bernhardt, 1997; Williams et al., 2002a; Weil, 2003]. A prevailing value of nondirective counseling is to acknowledge that the patient has the right to autonomous decision making that seems appropriate to the patient in view of her or his particular values, objectives, and ethical and religious standards [Ad Hoc Committee on Genetic Counseling, 1975; Kessler, 1979; Emery, 1984; Royal Commission, 1993; Andrews, 1997]. For example, the code of ethics for the National Society of Genetic Counselors states that counselors “should strive to enable their clients to make informed independent decisions, free of coercion, by providing or illuminating the necessary facts and clarifying the alternatives and anticipated consequences” [NSGC, 1992]. The value of patient autonomy is to be supported even where the health care professional personally may disagree with the patient’s decision (with regard to genetic testing of children under the age of 18 years for adult-onset disorders, professional organizations have stated that testing may be discouraged in absence of potential benefit; however, each case should be considered on an individual basis [Bloch and Hayden, 1990; Committee of IHA and WFN, 1990; Sharpe, 1993; Wertz et al., 1994; ASHG/ACMG Report, 1995; AMA,
2003; Bioethics Committee (CPS), 2003; WHO, 2003; but also see Duncan, 2004] (see also, Chapter 11, Susceptibility Testing).

However, as discussed in the following section by Nancy Callanan and Bonnie LeRoy, important distinctions are made between directing the process compared to the outcome of genetic counseling [Bartels et al., 1997; Kessler, 1997a; Biesecker, 2001], especially in those scenarios where patients may not have sufficient information [Bower et al., 2002], may perceive or retain information in a way not intended by the provider, and may wish to share the provider’s experiences and expertise [Royal Commission, 1993; Michie et al., 1997b, 1997c, 1998; Bernhardt et al., 2000; Bower et al., 2002]. The Canadian College for Medical Geneticists [CCMG, 2004b] professional accreditation standard for a clinical geneticist acknowledges these potential scenarios, stating that when providing genetic counseling:

*The physician should be able to be sympathetic and empathetic, remain objective and impartial, delineate his/her own ethical standards and appreciate those of the patient, appreciate the general mores of the culture of the patient, be non-directive in most instances, but be prepared to advise in certain situations and provide psychological support, either personally or through referral.*

With regard to outcomes, nondirectiveness may be found in conflict with those health care professionals who do not feel that medical decision making about outcomes should be the sole prerogative of the patient [Royal Commission, 1993; Geller et al., 1996; Quill and Brody, 1996; Andrews, 1997; see also section by Mykitiuk et al. in Chapter 8, Prenatal and Neonatal Screening]. Some have argued that nondirectiveness may be perceived as representing a neglect of the necessary role of accountability or responsibility in autonomous decision making [Yarborough et al., 1989; Pauker and Kassirer, 1987; Pauker and Pauker, 1987]. Although patient autonomy in decision making is acknowledged, the health care professional may have a significantly different perception from the patient about the impact of genetic disabilities [Royal Commission, 1993; Ormond et al., 2003] and may wish to make recommendations on the basis of those perceptions and experiences [Williams et al., 2002a; also see Geller et al., 1993; an American court has stated that where a patient selects a treatment from a group of reasonable alternatives and the physician regards the choice as inappropriate, the physician may withdraw from further care without liability for failure to provide treatment provided that the physician has reasonable assurances that treatment and care will continue: *McLaughlin v. Hellbusch*, 1999; see also section by Mykitiuk et al. in Chapter 8, Prenatal and Neonatal Screening]. For example, if a patient has been advised of a significant risk that her child will be born with Down syndrome, what course of action provides the most benefit and prevents harm? The health care professional may argue that the option with the greatest benefit will have the least burden [Pauker and Kassirer, 1987] such as the monetary cost of care and the avoidance of the birth of a genetically afflicted child [Pauker and Pauker, 1987]. If the patient, however, due to her attitudes toward Down syndrome decides to proceed with the pregnancy, the health care professional may perceive this to be the selection of the option with the lowest benefit. The patient’s choice, and underlying values and beliefs, therefore, may be deemed by the health care professional to be
Chapter 1 Genetic Counseling and the Physician–Patient Relationship

nondeliberative and irrational [Pitz, 1987]. Studies indicate, however, that the health care professionals may be equally susceptible to such “inconsistencies” [Pitz, 1987; Michie, 1997d; Michie and Marteau, 1999b]. One health care professional, on the basis of experience, may be of the opinion that a child afflicted with a genetic disorder can enjoy a full and satisfying life; another health care professional may believe that the financial and emotional costs of the same disorder justifies a recommendation of abortion [Abramsky et al., 2001].

Health care professionals who have not received specialized training in genetics, when confronted with situations where any course of action may result in harm to the patient (e.g., the decision whether to terminate a pregnancy or to give birth to a child with a severe genetic condition) may be uncertain as to the nature and extent of their roles and responsibilities [Veach, 2004]. How then, within the context of genetic testing, is medicine’s traditional ethical maxim—\textit{primum non nocere}, above all do no harm—to be defined and from whose perspective? For example, in prenatal diagnosis, no desirable option may exist, and any course of action, whether the termination of a pregnancy or the birth of an afflicted child, can result in human anguish and tragedy [Tymstra, 1991; Grant, 2000].

Martha Nussbaum, in \textit{The Fragility of Goodness: Luck and Ethics in Greek Tragedy and Philosophy}, discusses scenarios where any course of action can result in harm (Agamemnon was told by the gods that if his daughter was not offered for sacrifice, the expedition to Troy would be becalmed, and everyone, including his daughter, would die):

\textit{Agamemnon is allowed to choose; that is to say, he knows what he is doing; he is neither ignorant of the situation nor physically compelled; nothing forces him to choose one course of action over another. But he is under necessity in that his alternatives include no desirable options. There appears to be no incompatibility between choice and necessity here—unless one takes the ascription of choice to imply that the agent is free to do anything at all. On the contrary, the situation seems to describe precisely a kind of interaction between external constraint and personal choice that is found to one degree or another in any ordinary situation. For a choice is always a choice among possible alternatives; and it is a rare agent for whom everything is possible. The special agony of this situation is that none of the possibilities is even harmless [Nussbaum, 1986].}

The issue of choice with regard to outcomes in such clinical scenarios is not necessarily a matter of harm or benefit or what the physician may perceive to be “rational” or “irrational” decision making [Emery, 1984; Royal Commission, 1993; Andrews, 1997]. The issue is one of autonomy.

Rather than perceiving the physician–patient relationship as a paternalistic one that reflects an inherent disparity in knowledge and expertise [Katz, 1984], within the context of genetic counseling, the role of the health care professional is to weigh and balance personal observation, human and medical genetics knowledge, skills, and experience against available options and their respective risks and benefits. The patient’s role is to describe and evaluate symptoms, detail a family medical history,
and discuss expectations, objectives, concerns, beliefs, and values. The result of these respective contributions is an informed choice by the patient [Hartlaub et al., 1993; Sharpe, 1994a, 1997; Jonsen et al., 1998; but see Quill and Brody, 1996; see also section by Mykityuk et al. in Chapter 8, Prenatal and Neonatal Screening].

Nondirectiveness represents the acknowledgment of the complex and troubling challenges posed by the processes of genetic testing and counseling. Of practical importance is the fact that many geneticists and counselors subscribe to the associated values and objectives. As will be discussed, this lends both insight and guidance to the manner in which professional guidelines, professional accreditation practice norms, and specialty program content for genetic testing services are likely to be interpreted by geneticists, counselors, health care professionals, and a court of law.

Conflict in decision making is resolved through recognition of the patient’s right of autonomy in decision making, to choose the course of action that seems appropriate to the patient in view of his or her values, objectives, and ethical and religious standards [Ad Hoc Committee on Genetic Counseling, 1975; Kessler 1979; Emery, 1984; Royal Commission, 1993; Andrews, 1997]. It is the task of the health care professional and the patient to willingly remain exposed to such risks, for these are the risks inherent in any autonomous decision [Nussbaum, 1986].

WEB RESOURCES


Continuing Medical Education: Genetics

American Medical Association: Genetics in Clinical Practice: A Team Approach, an Interactive Medical Laboratory Virtual Clinic. This CD-ROM is now available at no cost to physicians and other health care professionals that have an interest in clinical genetics: http://www.ama-assn.org/ama/pub/article/1615-8311.html

National Coalition for Health Professional Education in Genetics: http://www.nchpeg.org


National Information Resources on Human Genetics and Ethics: http://www.georgetown.edu/research/nrcbl/nirehg/

Chapter 1  Genetic Counseling and the Physician–Patient Relationship

Genetic Counseling Organizations

American Board of Genetic Counseling: http://genetics.faseb.org/genetics/abgc_diplomates.html
National Society of Genetic Counselors: http://www.nsgc.org/resourcelink.asp
How to Find a Genetic Counselor (U.S.): http://www.genetichealth.com/Resources_How_to_Find_a_Genetic_Counselor.shtml
American Board of Genetic Counselors: http://genetics.faseb.org/genetics/abgc_diplomates.html
American Board of Genetic Counseling: http://www.abgc.net/
University of Kansas Medical Center: http://www.kumc.edu/gec/prof/gc.html

Glossary of Terms

University of Kansas Medical Center: http://www.kumc.edu/gec/glossnew.html
National Human Genome Research Institute: http://www.ornl.gov/sci/techresources/Human_Genome/glossary/
“Talking” Glossary of Genetic Terms: http://www.genome.gov/10002096

Laboratories

Association of Public Health Laboratories: Newborn Screening and Genetics: http://www.aphl.org/Newborn_Screening_Genetics/index.cfm
GeneTests: A publicly funded medical genetics information resource developed for physicians, other health care providers, and researchers, available at no cost to all interested persons: http://www.genetests.org/
Canadian College of Medical Geneticists: Cytogenetics: http://www.hrsrh.on.ca/genetics/CanCyt/
Directory of Medical Cytogenetic Laboratories in Canada: http://www.hrsrh.on.ca/genetics/canlabs.htm

Professional Organizations

American Board of Genetic Counselors: http://genetics.faseb.org/genetics/abgc_diplomates.html
American Board of Genetic Counseling: http://www.abgc.net/
American Board of Medical Genetics: http://www.abmg.org/
American College of Medical Genetics: http://www.acmg.net/
American Society of Human Genetics: http://genetics.faseb.org/genetics/ashg/ashgmenu.htm
Association of Genetic Technologists: http://www.agt-info.org/
Canadian College of Medical Genetics: http://ccmg.medical.org/
Council of Medical Genetics Organizations: http://genetics.faseb.org/genetics/ashg/comgo.htm
European Society of Human Genetics: http://www.eshg.org/
Genetics Society of America: http://www.genetics-gsa.org/
International Federation of Human Genetics Societies: http://www.ifhgs.org/
Latin American Network of Human Genetics Societies: http://www.relagh.ufrgs.br/
International Society for Nurses in Genetics: http://www.isong.org/
National Society of Genetic Counselors: http://www.nsgc.org/
National Coalition for Health Professional Education in Genetics: http://www.nchpeg.org/
National Society of Genetic Counselors: http://www.nsgc.org/resourcelink.asp
International Federation of Human Genetics Societies: http://genetics.faseb.org/genetics/ifhgs/
Coalition of State Genetics Coordinators (CSGC): http://www.stategeneticscoordinators.org/

GENETIC COUNSELING APPROACH TO GENETIC TESTING

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INTRODUCTION TO GENETIC COUNSELING

Historically, genetic counseling has been defined as a communication process that deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family [American Society of Human Genetics, 1975]. This often
Chapter 1 Genetic Counseling and the Physician–Patient Relationship

The quoted definition emphasizes the interactive nature of genetic counseling, as a process performed by appropriately trained individuals, which encompasses both education and counseling, and is performed in the context of respect for individual autonomy in patient decision making. Respect for individual patient autonomy continues to be a central guiding ethical principle in contemporary genetic counseling and is a basic tenet for the professional code of ethics adopted by the National Society of Genetic Counselors [NSGC, 1992]. The tradition of nondirectiveness as a means of respecting patient autonomy has formed the framework for the values of the profession and the practice of genetic counseling. The genetic counselor aims to appreciate the values of the patient and incorporate those values into the counseling and facilitated decision-making components that are vital to effective services.

In this chapter, we will present a brief overview of the history of the profession and discuss the evolving tradition of nondirectiveness. Some of the primary factors that have contributed to the shift in responsibility to primary health care providers for identifying patients at risk, providing genetics education, genetic testing, and referral to genetic specialists will also be explored. Additionally, the major categories of genetic testing and issues raised by each will be discussed.

HISTORICAL PERSPECTIVE AND THE ROLE OF NONDIRECTIVENESS IN GENETIC COUNSELING

Sorenson [1993] summarized the history of “genetic counseling” by describing three phases, with phase A representing the late 1800s to the late 1930s, phase B, the late 1930s to the late 1960s, and phase C the late 1960s and beyond. Phase A constituted the major period of eugenics in this country during which applied human genetics was practiced as part of a social movement. The belief that heredity contributed not only to medical but also to social conditions, such as poverty, crime, and mental illness, and enthusiasm over the possibility that genetics might be used to improve the human condition led, in the United States, to the development of institutions where data on human traits was collected, to state laws that mandated involuntary sterilization of individuals determined to be “mentally defective,” and federal laws to limit immigration by various “inferior” ethnic groups [Walker, 1998]. In phase B applied human genetics moved out of the organizational and institutional context of eugenics and into academia, practiced initially by Ph.D. academic-based geneticists, followed by the emergence of academic physician-geneticists, and ultimately to the establishment of clinical genetics departments of medical schools [Sorenson, 1993]. In 1947, the term genetic counseling was introduced by Sheldon Reed, who proposed that in order to be effective, genetic counselors must have the knowledge and ability to provide genetic information in the context of respect for the sensitivities, attitudes, and reactions of clients [Reed, 1955]. Recognizing the complexities of reproductive genetic decision making, and anxious to avoid an association with early eugenic programs, many clinical geneticists of the time also advocated a nondirective type of counseling [Resta, 1997]. Phase C was characterized by rapid advances in the field of human genetics, leading to further development of clinical genetics as a medical
specialty and the emergence of masters-level genetic counselors [Sorenson, 1993; Walker, 1998].

The value of informed reproductive decision making and nondirective counseling assumed major roles in the development of genetic counseling in the United States [Sorenson, 1993]. As the profession evolved, a nondirective counseling approach based on Carl Roger’s client-centered counseling model emerged as a good fit for genetic counseling [Fine, 1993]. Although the principle of respect for client autonomy remains a core value in the practice of genetic counseling, several have challenged the use of the term *nondirective* as it has been applied to the practice of genetic counseling. Narrow definitions of the term are problematic as they imply value neutrality that is both ethically insufficient and not achievable [Weil, 2003].

In describing nondirectiveness as “procedures aimed at promoting the autonomy and self-directedness of the client” Kessler [1997a] not only provided a more broad definition of nondirectiveness but also described the quality counseling skills that are needed in order to achieve this goal within the context of genetic counseling. A survey of genetic counselors [Bartels et al., 1997] was undertaken to assess how they defined nondirectiveness, its value to their practice, and whether there were circumstances in which they employed a directive approach. The results suggest that nondirectiveness is defined in a consistent way and is highly valued by genetic counselors; however, important distinctions between directing the *process* and the *outcome* of genetic counseling were apparent as respondents cited as examples of when a more directive approach was utilized. Clients rely on genetic counselors to share their expertise and experience and to provide guidance about the process. A totally value-neutral stance is neither achievable nor appropriate. Even within the context of nondirectiveness, it is appropriate for genetic counselors to fully utilize their knowledge of medical genetics, psychological, and ethical issues in the process of supporting client autonomy by promoting active, self-confident decision. Further work in describing an empirically established model that articulates the process of genetic counseling is still needed [McCarthy et al., 2002].

As primary health care providers are forced to address their patient’s questions about genetics, it is important to remain cognizant of the values central to the practice of genetic counseling. These values have guided the provision of service for more than 30 years and remain constant because they respect the ability of the patient to ultimately make best decision. Primary care providers must work in collaboration with genetic specialists to develop models for providing genetic risk assessment, patient education, and testing for their patients that incorporate the ethical principles that continue to serve as the underpinning of traditional genetic counseling.

**PRACTICE OF GENETIC COUNSELING TODAY**

The practice of genetic counseling has changed dramatically since its inception. The only tools available to genetic counselors years ago were the statistics drawn from observations of affected families that indicated the likelihood that someone might inherit or pass on a gene that could cause a disease. Direct gene testing did not exist.
Chapter 1 Genetic Counseling and the Physician–Patient Relationship

For instance, in reproductive genetic counseling, the only options available to most individuals at risk for having a child with a genetic birth defect involved decisions about whether or not to have children in light of the known risks and, after conception, whether to continue or terminate a pregnancy known to be or suspected to be affected. Today, couples may consider sophisticated prenatal testing and reproductive options such as egg donation, preimplantation genetic diagnosis, surrogate motherhood, and more. Advances in genetic research now allow for over 1000 tests available for diagnosis, risk assessment, prenatal, and predictive testing [GeneTests, 2004].

The popular media present information about new genetic tests and technologies to the public almost daily. Individuals and families are encouraged to seek genetic counseling and testing, and primary care providers are encouraged to offer genetic tests to their patients. Some laboratories are currently participating in direct-to-consumer marketing. These efforts are expected to result in an increased demand from patients for tests such as susceptibility testing for hereditary breast and ovarian cancer, the genes BRCA1 and 2. There are concerns that this type of marketing will adversely affect the care a patient receives in that patients may not be making fully informed decisions [Hull and Prasad, 2001a, 2001b]. The demand for genetic counseling services likely will increase in response to advances in genetics research. These advances are expected to lead to genetic population screening to better predict the risk for developing many common conditions such as diabetes, heart disease, and cancer [Collins and McKusick, 2001]. These are the disorders that will affect almost every family. These are also the disorders that result in the majority of health care costs today.

GENETIC COUNSELOR GOALS

Although there is a dearth of research investigating the model or models of practice for genetic counseling, the literature addressing this issue is relatively consistent with regards to genetic counselor goals. Studies suggest that giving information is not the only important factor in meeting the patient’s needs but that patients expect, welcome, and need supportive counseling at a genetic counseling visit. A study examining genetic counseling sessions [Matloff, 1994] found that the content of prenatal counseling sessions varied significantly among their sample. However, the majority of counselors listed patient education and informed decision making as their primary goals in the session. Bernhardt et al. [2000] interviewed 16 genetic counselors and 19 patients about genetic counseling goals. They identified the following as the major goals:

1. Increase in patient knowledge and understanding
2. Accurate risk assessment
3. Facilitated decision making
4. Patient support and anticipatory guidance
5. Alleviate guilt
6. Empower patients to feel in control
7. Make the necessary referrals
The patients in this study perceived the benefits of genetic counseling as including:

1. Acquisition of information and increased knowledge
2. Immediate psychosocial support
3. Long-term psychosocial support
4. Anticipatory guidance
5. Facilitation of family communication
6. Assistance with decision making

Both counselors and patients considered the nature of the interpersonal interaction of utmost importance. A survey [Lobb et al., 2001] of 29 Australian genetic counselors and clinical geneticists, working in cancer genetics, identified 5 counselor goals:

1. Assessing patient needs and concerns
2. Providing technical genetic information
3. Conducting an individual risk assessment in the context of supportive interaction
4. Discussing the pros and cons of genetic testing
5. Developing a follow-up plan

These findings may have limited applicability to U.S. genetic counseling, and they are further limited to one type of genetic counseling—cancer genetics, but working within the context of a supportive interaction was found to be a basic underlying goal. In a longitudinal study of 43 families who received genetic counseling, Skirton interviewed each family prior to counseling and twice after counseling, at 2 to 4 weeks and 6 months, respectively [Skirton, 2001]. The outcome of genetic counseling was influenced by four factors: the patients’ need for certainty, the quality of their relationships with the genetic counselor, the integration of lay and scientific explanations, and the patient’s psychological adaptation. The results of this study support the notion that genetic counselor behaviors play a major role in producing effective genetic counseling outcomes. Although all of these studies show variation in the way genetic counseling is practiced, the basic underlying goals are constant:

- An informed patient empowered to make autonomous decisions
- Respect for the values and decisions made by the patient
- Attending to psychosocial needs of the patient
- Providing supportive counseling and appropriate follow-up care

IMPORTANT CULTURAL CONSIDERATIONS IN GENETIC COUNSELING

Cultural differences often challenge a practitioner’s ability to provide effective medical care, but in the genetic counseling setting, these differences can become a true obstacle. In some cases, the history of genetics with respect to instances of blatant
discrimination remains in the collective memories of people whose ancestors were victims and impedes building the trust that is necessary to provide good medical care. Differences in language, ways of communicating, worldviews, and religious beliefs can hinder the process of communicating accurate information and examining the associated psychosocial issues that are fundamental to effective genetic counseling. In addition, there are often major differences in perceptions about the cause of a disorder and the consequent burden that that disorder places on the family. The genetic counselor cannot rely on her or his worldview for guidance. For example, a survey [Greeson et al., 2001] about the perception of disability in the Somali immigrant population in a city in the upper Midwest reported that participants were constant in their belief that disabilities and birth defects are considered a gift from God not to be questioned in their culture. Families who had a child with a birth defect were special because they were “chosen by God,” and God knew that they could care for this specific child. It was obvious that traditional genetic counseling would not be useful in this population. Practitioners would need to change their focus in order to be effective. Participants were very interested in learning about anything that would improve the health of a child (or any individual) but not in anything that would predict or prevent what God had planned [Greeson et al., 2001].

In many cultures, the traditional nondirective approach and concern for promoting patient autonomy may directly conflict with the expectation that health care providers will tell the patient what to do in order to best deal with the disease. Patients often have complete confidence that their health care provider will make the best decision for them, and, by not doing so, the patient may lose confidence in the abilities of health care provider. Lewis [2002] expresses concern about disparities in the genetic services received by members of culturally diverse groups, in particular, ethnic and racial minority groups. He refers to two genetic counseling models that have been proposed by Kessler [1997b]—a teaching model and a counseling model. Lewis states that although these models fail to explicitly address issues of culture, the counseling model may best support multicultural approaches to genetic counseling. Ota Wang [1993] argues that genetic counselors must develop a foundation of cultural knowledge, awareness of self and others, and specific counseling skills. These studies emphasize the importance of cultural influences in providing effective genetic counseling.

**FACTORS THAT PROMOTE EFFECTIVE GENETIC COUNSELING**

Although there are few studies examining what constitutes effective genetic counseling, attending to certain issues often contributes to a better outcome. Careful attention to patient expectations, perceptions, and needs is fundamental to effective genetic counseling. Many studies have examined patient expectations of genetic counseling, patient risk perception, motivations for wanting genetic counseling and genetic testing, and patient needs. However, these studies find that patients often do not know what to expect from genetic counseling or how to directly communicate their needs [Hallowell et al., 1997]. Most patients could benefit from precounseling education.
One strategy suggested in the literature for dealing with this issue is to send patients written information about genetic counseling prior to their appointment. If patients and families know what to expect, they will come to the session more prepared to communicate their needs. For many patients, the perceived burden of the disease is a major factor in the decision to seek genetic counseling and genetic testing. Many people have a difficult time relating risk to their own situation [Ravine et al., 1991]. Patients who are unaware of their risk or do not clearly comprehend their risk in relation to their own health usually do not see a benefit in genetic counseling services. When patients gain a better understanding of their risk and appreciate the options available to modify that risk, they are more likely to benefit from genetics services.

It is critical that the genetic counselor provide accurate and current information, but this is only one facet of effective genetic counseling. Equally essential for providing effective genetic counseling service is the counselor’s ability to ascertain the patient’s motivation for wanting testing. In some cases, the genetic test will not answer the patient’s questions or provide the patient with a level of certainty about the future. In other cases, patients may come in for testing in response to the urging of a third party such as a family member. Genetic counselors also need to explore the patient’s perception of risk and burden of the disease, assist the patient in identifying the specific issues that are important to their own health and family situation, and possess the skills needed to provide supportive counseling.

GENETICS IN MAINSTREAM MEDICAL CARE: THE EMERGING MILIEU

The steady surfacing of genetics in mainstream medicine has been recognized for quite some time. The National Coalition for Health Professional Education in Genetics (NCHPEG) was established in 1996 by the American Medical Association, the American Nurses Association, and the National Human Genome Research Institute. The major focus of NCHPEG is “a national effort to promote health professional education and access to information about advances in human genetics.” Over 100 health professional organizations, consumer and volunteer groups, government agencies, private industry, managed care organizations, and genetics professional societies are members of NCHPEG (http://www.nchpeg.org/). In 2001, NCHPEG published the document, Core Competencies in Genetics Essential for All Health-Care Professionals. This publication delineates the essential information needed by health care professionals as they encounter the integration of genetics into all aspects of medical practice.

Genetic counselors also are rapidly becoming practitioners in mainstream medicine. They now provide services in multiple settings at infertility clinics, cancer centers, psychiatric clinics, neurology and cardiology clinics, and many more. In such settings, patients requesting genetic counseling may be considering presymptomatic diagnosis for a later-onset condition such as Huntington disease or testing for predisposition to a familial cancer or more information about the role that inheritance
plays in heart disease. The near future of genetic counseling might involve gene testing to see if an existing condition, such as clinical depression, some forms of heart disease, or Alzheimer disease, has a genetic basis in a specific individual or family. Information from this type of testing likely will generate a more accurate risk assessment specific to an individual. The arena of pharmacogenetics will emerge as an increasing role in identifying genetic factors that affect response to drugs used for common ailments [Begley, 1999]. It will be possible to identify some individuals who are likely to respond poorly to a medication. These individuals may seek genetic counseling in some form to better understand the implications of these test results for themselves and other family members.

One of the major differences of genetic medicine when compared to traditional medical care is the impact that genetic information has on the whole family. Genetics means families and often the practitioner must work within the family system in order to be most helpful. When one individual is diagnosed with a genetic disease or found to carry a gene mutation, this information has direct implications for parents, brothers, sisters, children, and other family members. The strain on a marriage and the effects on family dynamics can be overwhelming. In a study looking at the effects of predictive testing for Huntington disease on marital relationships, researchers found that partners of at-risk individuals demonstrated significantly higher levels of depression than those individuals at risk [Quaid and Wesson, 1995]. A genetic condition has a wide range of affects on multiple family members and the resulting psychological repercussions often disrupt relationships. Exploring family issues with the patient prior to testing is a foremost element of successful genetic counseling.

Genetic information learned through a risk assessment and genetic testing can be devastating to an individual. The consequent emotional reactions often interfere with a complete understanding of the technical information needed by the patient to make decisions and cope with the disease. Common reactions include denial, anger, fear, despair, guilt and shame, sadness, and grief [Djurdjinovic, 1998]. Although these reactions occur when dealing with patients in many areas of medicine, guilt and shame play a particularly prominent role in genetic counseling. Patients may feel personally responsible for passing a gene on to a child or to be ashamed of the history of genetic disease in their family, believing that their family is defective. The diagnosis may mean a surprising and unwanted change in the patient’s life and affect the patient’s perception of self [Delaporte, 1996]. The reactions are often emotional and not cognitively based in fact. Once again these studies emphasize the importance of attending to the psychosocial needs of patients and families in the genetic counseling setting especially when the patient is considering a genetic test.

CATEGORIES OF GENETIC TESTING:
 ISSUES TO CONSIDER

The issues that surround genetic testing relate to the indication for doing the test. In considering these issues, genetic counselors distinguish genetic testing from genetic screening. The term genetic testing describes evaluation of individuals who have an
increased risk for a specific genetic condition based on family history or clinical symptoms. This can include diagnostic testing, or testing that is performed to confirm a clinical diagnosis of a genetic condition or syndrome, for example, performing a cytogenetic analysis to confirm the diagnosis of Down syndrome in an infant with features of this condition. As such, the use of cytogenetic or molecular genetic tests within the context of a diagnostic evaluation is not unlike other forms of laboratory testing.

Issues to consider in the use of diagnostic genetic testing include the following:

- The use of genetic testing to establish or confirm a diagnosis is consistent with providing the best medical care for patients. As with similar medical recommendations, a nondirective approach is not required.
- Diagnostic genetic testing of a patient often results in information concerning potential risks, either for the same diagnosis or for reproductive risks for other relatives. Therefore, it is important to discuss these potential implications prior to testing and to encourage patients to share relevant information with at-risk relatives. Strategies for providing information to patients and their relatives that is relevant and comprehensible must be employed.
- In some situations a health care provider may face situations in which a patient is unwilling to inform relatives about their genetic risks. The responsibility to protect the confidentiality of a patient’s medical information would prevent a health care provider from disclosing the information directly to at-risk relatives. However, in rare situations in which the risk is great, and effective medical interventions are available, it has been argued that limited disclosure may be acceptable [ASHG Social Issues Subcommittee on Familial Disclosure, 1998; Offit, 2004; also see Chapter 15, Confidentiality and Recall].

Genetic testing also is used to determine the genetic status of an individual who is at risk for carrying a gene mutation for a specific disease or genetic condition based on their family history, for example, a testing for cystic fibrosis (CF) mutations in a woman whose brother is affected with this condition. In this situation, carrier testing is accompanied by a genetic risk assessment based on pedigree analysis and should be preceded by a discussion of the risk, the availability of testing, the potential benefits and limitations of testing, and the implications of testing with regards to reproductive planning.

Issues to consider in the use of carrier testing of individuals with a positive family history for a specific disorder include:

- It is important to obtain accurate information about the family history as well as confirmation of the diagnosis in the affected relative prior to testing [Uhlmann, 1998].
- Accurate testing may require information about the specific genetic mutation identified in the affected relative. Therefore, cooperation of relatives is needed in order to provide accurate testing for your patient. If such information is not available, then the limitations of testing must be communicated to the patient.
Chapter 1 Genetic Counseling and the Physician–Patient Relationship

- Carrier testing should be preceded by a discussion of the genetic risk, as well as the potential implications of the test result, including reproductive risks and options.
- Counseling strategies should be employed to assist patients in evaluating the pros and cons of testing in their situation in light of their own values, beliefs, and goals [McCarthy Veach et al., 2003].
- Carrier testing for familial genetic conditions in minors is generally discouraged [ASHG/ACMG, 1995; Davis, 1998].

Prenatal diagnosis is genetic testing performed during a pregnancy for individuals who are at risk for a specific condition due to family history, known carrier status, or prior reproductive history, for example, using amniocentesis to obtain a sample for biochemical or molecular testing for Hurler syndrome in a woman whose first child was affected by this disorder.

Issues to consider in the use of prenatal genetic diagnosis include:

- A careful assessment of the reproductive risks, availability of testing, evaluation of the accuracy, sensitivity, and potential limitations of testing should also precede prenatal diagnosis [Walker, 1996].
- Informed consent also should be obtained prior to prenatal diagnostic testing. This should include a full discussion of any risks to the procedures as well as the limitations of testing.
- Genetic counseling strategies that support patient autonomy and facilitate patient decision making are crucial prior to prenatal diagnosis [McCarthy Veach et al., 2003].

The term genetic screening is used to describe testing for genetic disorders in specific groups of people independent of a family history or clinical features. Criteria for genetic screening can include factors such as age, gender, or ethnic background [McCabe and McCabe, 2002]. Examples of genetic screening include newborn screening for the diagnosis for conditions such as phenylketonuria (PKU), prenatal screening by evaluation of maternal serum markers for open neural tube defects, Down syndrome and other chromosome disorders, and carrier screening for conditions such as cystic fibrosis or Tay-Sachs disease in individuals who belong to populations known to be at increased risk for these disorders. Offering prenatal testing for Down syndrome and other chromosome disorders to women over the age of 35 is also a form of genetic screening. It is anticipated that genetic screening for common disorders, such as Type II diabetes will be developed within the predictable future [Khoury et al., 2003].

Issues to consider in the use of genetic screening tests include:

- It is important to educate patients about the difference between screening tests and diagnostic tests as well as the possibility of false-negative and false-positive results.
Categories of Genetic Testing: Issues to Consider

- Participation in genetic screening programs should be voluntary and accompanied by education and informed consent prior to screening. Individuals considering screening tests should be informed about the potential risks of screening, which include the possibility for stigmatization and for genetic discrimination in life or health insurance and/or employment.

- Screening programs should make genetic counseling available to individuals who are diagnosed with or found to be carriers of genetic disorders or susceptibilities.

- As with diagnostic genetic testing, the results of genetic screening tests frequently have implications for other relatives.

The term *predictive genetic testing* is used to describe genetic testing in healthy individuals for genetic disorders with late onset. This includes *presymptomatic testing*, for example, testing for Huntington disease in individuals at risk for this condition prior to the onset of symptoms. It also includes *susceptibility testing*, or testing for genetic changes that predispose an individual for certain health conditions. Testing for *BRCA1* or *BRCA2* mutations in a woman with a family history of breast cancer is an example of susceptibility testing. The distinction between presymptomatic and susceptibility testing is important because the former implies certainty of developing the specific condition at some point in the future, while the latter identifies individuals who have an increased risk for developing a specific condition.

Issues to consider in the use of predictive genetic testing include:

- Careful psychosocial assessment and exploration of the reasons for seeking testing should precede any type of predictive testing. Alternatives to testing should be discussed, and counseling strategies to help patients evaluate the pros and cons of testing in the context of their own values, needs, and beliefs should be employed [Armstrong et al., 2002].

- Follow-up with regard to medical management issues and assessment of the patient’s emotional response to testing is important in any type of predictive testing, regardless of the results.

- Testing of minors for adult-onset conditions is generally discouraged, with the exception of situations in which there is a clear medical benefit to testing [ASHG/ACMG, 1995]. Counseling strategies to help parents explore their rationale for seeking predictive testing in their children is important.

- As with any type of genetic testing, confidentiality of test results is required. However, the results of testing in one patient often have implications for other relatives. Patients should be encouraged to share information about potential risks with their relatives, and health care professionals should be prepared to facilitate this process.

- It is important to note the difference between predictive testing and susceptibility testing. Although there is a lot of overlap of the issues to consider for both types of testing, it is especially important in susceptibility testing to ensure that patients are aware of the limitations of the test results.
As illustrated in the examples provided above, there are important issues that surround all types of genetic testing and screening. These relate directly to the core values and goals of genetic counseling: promoting patient autonomy by providing information, sharing expertise and support, and employing counseling strategies to facilitate decisions that reflect the patient’s own values and beliefs. While there is considerable overlap in issues, by focusing on the specific context in which testing is offered or performed, the practitioner can be better prepared to anticipate issues and approach patients appropriately.

SUMMARY

Genetic counseling is an exceptionally complicated process that is fundamentally different from other areas of medical practice in that it is almost always necessary to consider not only the needs of the patient but the impact on the patient’s family or prospective family. Moreover, deep-seated personal and cultural values influence the decisions patients make about genetic testing. Many genetic tests generate information that changes self-perception, alters family relationships, and compels patients to adjust their life choices. Given the potential for serious consequences resulting from genetic information and genetic testing, it is essential that the provider possess the skills needed to address all of these issues with patients and help patients find the decision that is right in their situation.

Genetic counseling has supported a nondirective counseling approach as a means of promoting patient autonomous decision making. Nondirectiveness has been a way of thinking about an approach that centers the counseling on the values of the patient and not those of the counselor. The profession of genetic counseling has evolved over the past 30 years in response to the extensive developments in our understanding of the role of genetics in disease and the increase in available technologies. With this evolution, challenges to the actual significance of the term nondirectiveness have emerged. Genetic counselors now have a history of practice such that practitioners are able to share their wealth of experience and recognize that appropriate professional advice and guidance is not synonymous with coercion. Nonetheless, the core goals of striving to understand the world of the patient as he or she sees and experiences it and of focusing decisions around the values of the patient remains central to genetic counseling. Over the years, Dr. Seymour Kessler has constantly reminded us of what is most important in the practice of genetic counseling through his multitude of publications and invited speaking engagements. Over and over he tells us that the critical question is not, “Am I being directive with this patient?” but rather, “How can I best help this patient.” Resta and Kessler [2004] summarize the basic counseling skills required to provide good genetic counseling:

- The ability to understand the psychological needs of others
- The ability to understand the psychological meaning of client’s behaviors
- The ability to communicate that understanding in ways that leave clients emotionally enriched, psychologically stronger, and more competent to deal with their own lives
We still have much to learn about the ways in which advances in genetics is impacting traditional medical practice. Moreover, it continues to be important to study the means by which patients and their families cope with genetic information and how all health care practitioners can best help their patients. Nonetheless, it is obvious that the genetic counselor in practice today needs to not only know how to obtain, comprehend, and communicate the most current technical information about the disease affecting a patient and/or their family but, in addition, possess the ability to empathize with patient’s situation, facilitate informed decisions, and help patients and families cope with their lives. As other health care professions enter the arena of providing genetic information to patients, they need to hold themselves to these same standards.