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The Practice of Genetic Counseling

Ann P. Walker, M.A., C.G.C.

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THE PRACTICE OF GENETIC COUNSELING

Historical Overview

Until the beginning of the last century there existed little scientifically based information for those concerned about the chances of an apparently familial disorder or birth defect occurring (or recurring) in themselves or their offspring. Observations of such conditions had sometimes led to correct interpretations of their pattern of inheritance, as in the understanding of hemophilia evidenced by the Talmudic proscription against circumcising brothers of bleeders, in Broca's report of a seemingly dominant breast cancer predisposition in five generations of his wife's family (Broca, 1866), or in societal taboos against marriages between close relatives. Often, however, birth defects and familial disorders were attributed to exogenous causes—punishment (or perhaps, favor) by a deity, a misdeed on the part of the parents (usually the mother), a fright, a curse, or some natural phenomenon such as an eclipse. Indeed, similar beliefs are still widespread in many cultures and may even figure subliminally in irrational fears of people who are otherwise quite scientifically and medically sophisticated.

Throughout the late 1700s and the 1800s, investigators wrestled with how traits might be transmitted. Lamarck's theories regarding the inheritance of *acquired*

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characteristics persisted into the twentieth century. Darwin recognized that characteristics that were advantageous in particular circumstances might increase the likelihood of survival and reproduction—eventually generating a population sufficiently different from its ancestors as to constitute a new species. Darwin's cousin Galton, by studying families and twin pairs, attempted to develop mathematical models to tease out the relative contributions of environment and heredity. By the start of the twentieth century, Bateson and Garrod had each recognized that the familial occurrence of alcaptonuria (described by Garrod in 1899) and other recessive "inborn errors of metabolism" could be explained by the neglected and recently rediscovered laws of Mendel (Garrod, 1902). Thus began a new era in which the pattern of inheritance of certain genetic conditions—and hence their risks of recurrence—could be deduced, providing a more scientific basis for genetic counseling.

During the last century, understanding of genetic disorders, variability, mechanisms, and contributions to common diseases grew exponentially. Medical technology exploded, leading to a host of new genetic testing capabilities, including prenatal and ultimately preimplantation genetic diagnosis. Less dramatic but equally important advances occurred in the study of human behavior, in public health policy, in ethics, and in counseling theory. Concomitantly, people began to assume greater responsibility for their own health care decisions. The *activity* of genetic counseling developed and changed accordingly over this period. It is only since the 1970s, however, that a *profession* specifically devoted to genetic counseling has arisen. The education and practice of these professionals encompasses all of the above elements, enabling them, as members of genetics health care teams, to bridge such diverse disciplines as research scientist, clinical geneticist, primary health care provider, social worker, and hospital administrator. More importantly, today's genetic counselor provides a service that is unique—distinct from the contributions of these other individuals—for patients and families who seek to understand and cope with both the genetic and the psychosocial aspects of disorders they confront.

Less than 40 years after the first master's degrees were awarded in genetic counseling, these new professionals have achieved a prominent place in genetic health care delivery, education, and public policy development. They have formed professional organizations in several countries, been involved in starting training programs, developed mechanisms for accrediting over 30 North American genetic counseling graduate programs, and become board certified, credentialed, registered, and/or licensed as distinct health professionals. This chapter gives an overview of these developments—and perhaps also a glimpse of the challenges and excitement to come.

Models of Genetic Counseling

Eugenic Model Sheldon Reed is credited with introducing the term "genetic counseling" in 1947 (Reed, 1955). However, the practice of advising people about inherited traits had actually begun about 1906, shortly after Bateson suggested that the new medical and biological study of heredity be called "genetics." By then the

public (and many scientists) had been intrigued by the thought that this new science might be able to identify hereditary factors contributing not only to medical diseases, including mental retardation, but also to social and behavioral diseases such as poverty, crime, and mental illness. Galton himself had suggested in 1885 that "eugenics" (a word he coined from the Greek ευγενηζ, meaning "well-born") become the study of "agencies under social control that may improve or impair racial qualities of future generations, either physically or mentally" (Carr-Saunders, 1929).

Enthusiasm over the possibility that genetics might be used to improve the human condition gave rise, for example, to the Eugenics Records Office at Cold Springs Harbor (a section of the Carnegie Institution of Washington's Department of Genetics) and establishment of a chair of eugenics (by bequest of Galton himself) at University College London. Not only did scientists in these institutions collect data on human traits, they also sometimes provided information to affected families—usually with the intention of persuading them not to reproduce. Unfortunately, at least at the Eugenics Records Office, data collection was often scientifically unsound, or was biased and tainted by social or political agendas. The eugenics movement, initially well-intentioned, ultimately had disastrous consequences. By 1926, 23 of the 48 United States had laws mandating sterilization of the "mentally defective" and over 6000 people had been sterilized (most involuntarily) (Carr-Saunders, 1929). Astoundingly, this practice persisted up into the 1960s and 1970s in some countries (Wooldridge, 1997). In 1924 the U.S. passed the Immigration Restriction Act, instituting quotas to limit immigration by various "inferior" ethnic groups. In Germany, euthanasia for the "genetically defective" was legalized in 1939—leading to the deaths of over 70,000 people with hereditary disorders in addition to Jews, Romanies (gypsies) and others killed in the holocaust (Neel, 1994). Revulsion at the specter of these past abuses in the name of mandatory eugenics is at the heart of the "nondirective" approach to genetic counseling that prevails today.[1]

Medical/Preventive Model Distress at the outcomes of what had started out as legitimate scientific inquiry caused most geneticists to retreat from advising families about potentially hereditary conditions for at least a decade. However, by the mid-1940s, heredity clinics had been started at the Universities of Michigan and Minnesota and at the Hospital for Sick Children in London (Harper, 2004). A decade later, during a time when prevention had become a new focus of medicine, several additional genetics clinics were established. Information about risks was offered—based almost entirely on empirical observations—so that families could avoid recurrences of disorders that had already occurred. However in 1956, few diagnostic tests were available. Knowledge of the physical structure of DNA was only three years old; there was no way to prospectively identify unaffected *carriers* of genetic conditions; and given that it was still thought that there were 48 chromosomes in the human genome and that our mechanism of sex determination was the same as in *Drosophila* (Therman, 1993; Miller and Therman, 2001), the basis for chromosomal

[1] Robert G. Resta has written an excellent essay reviewing the complex issues around eugenics and nondirectiveness for the *Journal of Genetic Counseling* (1997, 6:255–258).

syndromes was completely unknown. Even with the goal of preventing genetic disorders, there was little for genetic counseling to offer families but information, sympathy, and the option to avoid childbearing. Many geneticists assumed that "rational" families would want to do so (Resta, 1997).

Decision-Making Model The capabilities of genetics changed dramatically over the next 10 years as the correct human diploid complement of 46 was reported by Tjio and Levan (1956) and the cytogenetics of Down (Lejeune et al., 1959), Klinefelter (Jacobs and Strong, 1959), and Turner (Ford et al., 1959) syndromes and trisomies 13 (Patau et al., 1960) and 18 (Edwards et al., 1960; Patau et al., 1960; Smith et al., 1960) were elucidated. Over this decade it also became possible to identify carriers for α- or β-thalassemia (Kunkel et al., 1957; Weatherall, 1963), a host of abnormal hemoglobins, and metabolic diseases such as galactosemia (Hsia, 1958), Tay–Sachs disease (Volk et al., 1964), and G6PD deficiency (Childs, 1958), among others. Amniocentesis was first utilized for prenatal diagnosis—initially for sex determination by Barr body analysis (Serr et al., 1955)—and then for karyotyping (Steele and Breg, 1966). In 1967, the first diagnosis of a fetal chromosome anomaly was reported (Jacobson and Barter, 1967).

These advances in genetics meant that families had some new options to better assess their risks and possibly avoid a genetic disorder. However, the choices were by no means straightforward. Tests were not always informative. Prenatal diagnosis was novel, and its potential pitfalls were incompletely understood. Explaining the technologies and the choices was time-consuming. However, clinical genetics' tenet of nondirective counseling was beginning to be echoed elsewhere as medicine began to shift from its traditional, paternalistic approach toward promoting patient autonomy in decision-making. The emphasis in genetic counseling shifted too, from simply providing information that families would presumably use to make "rational" decisions (thereby preventing genetic disorders) toward a more interactive process in which individuals were not only *educated* about risks but also helped with the difficult tasks of exploring issues related to the disorder in question, and of making decisions about reproduction, testing, or management that were consistent with their own needs and values.

Psychotherapeutic Model Although families often come to genetic counseling seeking information, they cannot really process or act on it effectively without dealing with the powerful reactions this information can evoke. For this reason, exploring with clients their experiences, emotional responses, goals, cultural and religious beliefs, financial and social resources, family and interpersonal dynamics, and coping styles has become an integral part of the genetic counseling process. Genetic disorders and birth defects often catch individuals completely off-guard—raising anxiety about the unfamiliar, assaulting the self-image, provoking fears for one's own future and that of other family members, and generating guilt. Even a client who brings a lifetime of experience with a disorder, or who has known about his or her own or reproductive risk for some time, will have cognitive or emotional "baggage" that may need to be addressed for counseling to succeed. A skilled genetic counselor must be able

to elicit and recognize these factors, distinguish appropriate from pathological responses, reassure clients (when appropriate) that their reactions are normal, prepare them for new issues and emotions that may loom ahead, and help them marshal intrinsic and extrinsic resources to promote coping and adjustment. A few genetic counselors have chosen to develop these skills to a higher degree by obtaining additional training so that they can provide longer-term therapy for dysfunctional families or for individuals whose underlying psychopathology complicates genetic counseling.

DEFINITION AND GOALS OF GENETIC COUNSELING

1975 ASHG Definition of Genetic Counseling

In the early 1970s a committee of the American Society of Human Genetics (ASHG) proposed a definition of genetic counseling that was adopted by the Society in 1975. Though oft cited, no textbook of genetic counseling would be complete without it:

> Genetic counseling is a communication process which deals with the human problems associated with the occurrence or risk of occurrence of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or 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) choose a course of action which seems to them appropriate in their view of their risk, their family goals, and their ethical and religious standards and act in accordance with that decision, and (5) to make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.
>
> —American Society of Human Genetics, 1975

This definition held up quite well for a time, articulating as it does several central features of genetic counseling. The first is the two-way nature of the interaction—quite different from the "advice-giving" of the eugenics period or the primarily information-based counseling characteristic of the mid-twentieth century. The second is that genetic counseling is a *process,* ideally taking place over a period of time so the client can gradually assimilate complex or distressing information regarding diagnosis, prognosis, and risk and formulate decisions or strategies. The third is the emphasis on the client's autonomy in decision-making related to reproduction, testing, or treatment, and the recognition that such decisions will *appropriately* be different depending on the personal, family, and cultural contexts in which they are made. The fourth acknowledges that the occurrence or risk for a genetic disorder can have a family-wide impact different from that in other kinds of diseases and indicates that there should be a psychotherapeutic component of genetic counseling to help people explore and cope with the reproductive implications and other burdens of a rare disorder. Implicit in the words "appropriately trained persons" is the admonition that, because of these particular features, genetic counseling requires special knowledge and skills distinct from those needed in other medical and counseling interactions.

Genetic Counseling Has Changed Since 1975!

More Indications for Genetics Services and Counseling The ASHG definition relates primarily to genetic counseling in the context of *reproductive* risk assessment and decision-making. In the three-plus decades since this definition was proposed, genetic counseling's purview has expanded considerably beyond the prenatal and pediatric realm, with many genetic counselors now focusing entirely on diagnosis and risk assessment for diseases that affect individuals as adults—frequently *after* they have completed their reproductive years. Moreover, genetic counseling often addresses conditions that are not solely, and sometimes not at all, genetic. Genetic counselors now provide information about potentially teratogenic or mutagenic exposures; about birth defects that may have little if any genetic basis; and about common diseases of adulthood that have complex and heterogeneous causes. Increasingly, counselors work in settings where they are involved in discussions about possible interventions like chemoprevention, prophylactic surgery, or other strategies, enabling patients to make choices that may reduce future disease risk. It is likely that in the future, as our understanding of the genome enables us to personalize medicine to individual genotypes, genetic counselors will have a role in discussing genetic polymorphisms that could affect a patient's response to therapeutic drugs or environmental pollutants or perhaps even in providing information about genetic variations that contribute to common behavioral and physical traits.

Changes in Clients and Health Care Delivery As individuals seeking genetic counseling have become more diverse and the technology ever more powerful and complex, new elements have gained prominence in the genetic counseling process. In 1975 one could not have predicted that access to genetic evaluation or appropriate treatment would be limited by lack of insurance or by constraints imposed by managed care, with the result that advocating for funding would become a new (and usually unwelcome) part of the genetic counseling process. Or that the counselor would need to inform clients not only about the nature of the disorder, risks, testing, and reproductive options, but also about ethical dilemmas that might arise as a result of testing, or about the possibility of resultant discrimination in employment or insurance. Or that the genetic counseling "process" might have to be accomplished in just half an hour. Or that counseling a recently arrived immigrant might be severely compromised by passage through two translations or the client's unfamiliarity with even rudimentary concepts of biology. The basic tenets and goals remain as they were in 1975, but the face of genetic counseling will continue to change.

2006 NSGC Definition of Genetic Counseling and 2007 Scope of Practice

Because genetic counseling has continued to evolve, in 2003 the National Society of Genetic Counselors (NSGC) appointed a task force to revisit the definition of genetic counseling. Recognizing that many types of professionals provide genetic counseling, the group's charge was to define *genetic counseling*, rather than to describe various

professional roles of genetic counselors (Resta et al., 2006). In reviewing the literature, the task force found 20 previous definitions of genetic counseling, which are provided as an appendix to their article. They also considered the purposes for which a genetic counseling definition might be used. Among these are marketing the profession, not only to potential clients but also to insurance companies, hospital administrators, and health maintenance organizations; increasing public, professional, and media awareness; developing practice guidelines and legislation for licensure; and providing a basis for research in genetic counseling. They settled on crafting a succinct definition that would be readily understandable, broad enough to apply to the variety of settings in which genetic counseling might occur, and acknowledging the increasing importance of genetic counseling for common and complex diseases. Drafts were presented to the NSGC membership and an advisory expert panel, as well as to representatives of other professional genetics organizations, advocacy groups, legal counsel, and marketing consultants for comments, and revised repeatedly to reflect this input. As approved by the NSGC Board of Directors, the definition reads:

> Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease. This process integrates the following:
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> - Interpretation of family and medical histories to assess the chance of disease occurrence or recurrence.
> - Education about inheritance, testing, management, prevention, resources and research.
> - Counseling to promote informed choices and adaptation to the risk or condition.

Note that the definition does not indicate who is qualified to *provide* genetic counseling. Nor does it address the scope of practice of genetic counselors. A second NSGC task force developed a complementary document to define genetic counselors' scope of practice and to capture the broad range of activities involved in genetic counseling.

"Scope of practice" is a term frequently used in the context of licensing non-physician medical professionals—particularly those with advanced practice degrees such as physician assistants, nurse practitioners, audiologists, etc. A scope of practice describes activities that an appropriately trained and qualified person in that profession should be able (and allowed) to do. It is usually developed by one or more organizations representing the profession as a means of educating others about their training, skills, and unique place in service delivery, and for encoding in regulatory language the tasks a licensed professional should be entitled to perform. Sometimes the scopes of practice of different professional groups overlap—occasionally causing tension if a newer professional group begins to provide services that historically have been the sole province of another. Since clinical geneticists and genetic counselors have practiced as a team since the beginning, this has been less of an issue in the provision of genetics services.

Describing a scope of practice is often viewed as an important step in a profession's development. While this was part of the reason the NSGC undertook the task, a more urgent reason was to provide a document that could be used in efforts to educate legislators about the need for genetic counselor licensure and to assist states in developing licensure regulations that would be as uniform as possible. The NSGC Scope of Practice describes elements of the genetic counselor's role as they relate to clinical genetics, to counseling and communication, and to professional ethics and values. The 2007 Scope of Practice is reproduced in the Appendix to this chapter, after the ABGC competencies. Because of the changing face of genetic counseling and its many potential future directions, it is anticipated that the Scope of Practice will be revised periodically to reflect how the profession is changing. The NSGC website should be accessed for the most up-to-date version.

Philosophy and Ethos of Genetics Services and Counseling

Voluntary Utilization of Services Genetic counseling operates on a number of assumptions or principles. Among these are that the decision to utilize genetics services should be entirely voluntary. Society at large and other entities such as insurance companies clearly have economic and eugenic interests in promoting prevention of genetic disease. However, at least in most developed nations, the prevailing philosophy is that information should be made available and tests offered when appropriate, but that patients and families should have the right to make their own decisions—particularly about genetic testing and reproduction—unencumbered by pressure or by the implication that they are being fiscally or socially irresponsible if they choose *not* to try and prevent a hereditary disease.

In reality, of course, patients sometimes get referred for genetics services not at their own request, but by virtue of a care provider's fear of litigation, or because they have been identified through a screening program about which they were not adequately educated. Furthermore, decisions about testing or reproduction are often influenced by financial considerations. Genetic disorders usually come with additional health care costs, which may or may not be covered by health insurance or public medical assistance programs. In some cases, insurers consider newer genetic tests to be "experimental" or see genetic counseling as unnecessary outside of the context of pregnancy. To assume that families can always make voluntary decisions about utilizing genetic services or about reproduction based solely on their preferences, personal goals, and moral views is, unhappily, somewhat naive. To maximize the ability of families to benefit from advances in genetics, it is incumbent on genetic counselors to educate insurers about the value of genetics services and testing, to advocate for access to these services, and also to be involved in developing public policies that promote responsible use of genetics, ensure that patients will be able to *make* choices, and also protect them from misuse of genetic information.

Equal Access Ideally, genetics services, including counseling, diagnosis, and treatment should be equally and readily available to all who need and choose to use them. Compared with other medical specialties, however, *genetics* services are more likely to be accessed by people living in heavily populated areas who have

some sort of health coverage, enough education or medical sophistication to know that such services exist, and the ability to advocate for themselves in the health care system. Even patients lucky enough to have these attributes may find particular genetic technologies (e.g., preimplantation diagnosis) out of reach, just because of their cost, novelty, or limited availability. As capabilities continue to expand it will be important to assess expensive genetic services not only in terms of how likely they are to be available to *all* those who might benefit, but also by considering their costs relative to other public health care needs.

Client Education A central feature of genetic counseling is a firm belief in the importance of client education. Expanding on the NSGC definition, this education typically includes information about (1) the features, natural history, and range of variability of the condition in question, (2) its genetic (or nongenetic) basis, (3) how it can be diagnosed and managed, (4) the chances it will occur or recur in various family members, (5) the economic, social, and psychological impact—positive as well as negative—that it may have, (6) resources available to help families deal with the challenges it presents, (7) strategies that can ameliorate or prevent it if the family so wishes, and (8) relevant research that may contribute to understanding the disorder or better treatment.

Complete Disclosure of Information In providing education about diagnosis and related issues, most geneticists and genetic counselors subscribe to the belief that all relevant information should be disclosed. Being selective in what one tells a client is viewed as paternalistic—and disrespectful of the person's autonomy and competence. There is wide disagreement, however, both in philosophy and in practice, on what geneticists or counselors view as "relevant". Most would probably concur that a competent patient should be given the facts about his or her own diagnosis—even in a challenging scenario such as informing a phenotypic female with androgen insensitivity syndrome about her XY karyotype. But there is less consensus about what should be done with other dilemmas, like disclosing nonpaternity revealed through DNA testing when it does not affect risk. With genetic testing now widely available for a host of carrier states, there also has been considerable debate about whether all possible tests (e.g., for diseases more prevalent in specific populations but still quite uncommon) need to be discussed or offered. Nor is it clear whether a counselor should be obligated to address issues of potential genetic significance that are not related to the reason for referral (e.g., a familial cancer history uncovered in the context of prenatal diagnosis counseling).

At the same time as testing capabilities and understanding of genetic mechanisms have become more extensive and complex, clients have become more diverse in their cultural backgrounds, education, and health literacy. Concomitantly, the time available for counseling has often decreased. In the 1980s and early 1990s, when a typical session might last an hour and a half and the majority of clients were college educated, middle class, and English speaking, a "genetics lesson" was a prominent feature of genetic counseling. We believed that clients needed a basic understanding of genes, chromosomes, and how the test would be done in order to make informed decisions. Now, however, with burgeoning genetic knowledge and technology, the pressure to

see more clients more quickly, and a more frequent need to work through interpreters, achieving this level of client education is often impractical. Moreover, full disclosure of all "relevant information" could paralyze even the most sophisticated patient. Despite these pressures, however, it will always be critical for the counselor to disclose any information *relevant to decision-making* in ways that the client can interpret and act on.

Nondirective Counseling Although the counselor can use clinical judgment in choosing what information is most likely to be important and helpful in a client's adjustment to a diagnosis or for decision-making, it should be presented fairly and even-handedly—not with the purpose of encouraging a particular course of action. Adherence to a nonprescriptive (often less appropriately referred to as "nondirective") approach is perhaps the most defining feature of genetic counseling. The philosophy stems from a firm belief that genetic counseling should, insofar as is possible, be devoid of eugenic motivation. Although this is a time-honored tradition, it can be counter-productive for the counselor to avoid expressing *any* opinions. This is especially true when genetics evaluation reveals *personal* health risks, such as an increased liability to specific diseases that could be reduced by particular interventions (e.g., aggressive screening, chemoprevention, or prophylactic surgery to reduce breast or ovarian cancer risk in a *BRCA* mutation carrier; monitoring serum iron, dietary modifications, or therapeutic phlebotomy in a person with hemochromatosis). It is even true in certain situations involving reproductive decision-making—an area where genetic counselors have historically shied away from expressing opinions or offering advice. If a pregnancy risk could be reduced by various actions (e.g., avoiding exposure to a teratogenic drug, taking folate, or achieving good diabetic control before to pregnancy), few counselors would hesitate to advise the client accordingly. A client should expect a genetics professional to be able to provide guidance when the genetic and medical issues are complex, if there are limited data or medical opinions conflict, and even when choices raise problematic moral or psychosocial issues. Failing to share our knowledge and experience out of fear that we will be perceived as directive may leave the client to flounder.[2]

Attention to Psychosocial and Affective Dimensions in Counseling Just giving information does not necessarily promote client autonomy. To succeed in empowering individuals to cope with a genetic condition or risk, and to make difficult decisions with which they are comfortable, the counselor needs to

[2] Seymour Kessler has explored nondirectiveness in genetic counseling—particularly in the context of reproductive genetics—in numerous publications, but the following is especially provocative: Kessler (1997) Psychological aspects of genetic counseling. XI. Nondirectiveness revisited. *American Journal of Medical Genetics* 72:164–171. Some more recent discussions of this challenging issue can be found in Elwyn, Gray, Clarke (2000) Shared decision making and non-directiveness in genetic counselling. *Journal of Medical Genetics* 37:135–138; in Oduncu (2002) The role of non-directiveness in genetic counseling. *Medicine, Health Care, and Philosophy* 5:53–63; and in Koch, Svendsen (2005) Providing solutions—defining problems: the imperative of disease prevention in genetic counselling. *Social Science & Medicine* 60:823–832.

encourage clients to see themselves as competent and to help them project how various events or courses of action could affect them and their family. This cannot be done without knowing something of their social, cultural, educational, economic, emotional, and experiential circumstances. The client's ability to hear, understand, interpret and utilize information will be influenced by all of these factors. An effective counselor will be attuned and responsive to affective responses and able to explore not only clients understanding of information, but also what it means to them, and what impact they feel it will have within their social and psychological framework.

Confidentiality and Protection of Privacy Respecting confidentiality and protecting personal health information has always been an essential part of any medical interaction, but it has become even more critical since passage of The Health Insurance Privacy and Accountability Act (HIPAA) in 1996. However, genetic counseling raises additional issues with regard to confidentiality and privacy protection. Information about an individual's genetic disease, family history, carrier status, reproductive risk, or related medical decisions is extremely sensitive and potentially stigmatizing. Very rarely, information about risk leads to discrimination in employment or difficulties in obtaining or retaining insurance. For these reasons it is especially critical that genetic information be kept confidential. On the other hand, knowing a person's diagnosis or genotype sometimes provides information not only about his *own* risk, but also that of family members who may be only remotely related. This can create a conflict between the client's right to privacy and the benefit to relatives of knowing about their potential risk. If the risk is substantial or serious, and when options are available to prevent harm, the client—and sometimes the counselor—may have an ethical duty to warn relatives. There are only a few other situations in medicine (e.g., a serious infectious disease or threat to another's safety disclosed in the course of psychotherapy) where breaching confidentiality may be warranted if the client refuses to share information with those at risk (please see Chapter 12 by Schmerler in this book for additional discussion).

With the advent of computerized databanks and of samples containing DNA being stored for many reasons, concerns have been raised about the *privacy* of genetic information. Genetic material obtained for one purpose (for instance, genetic linkage studies, newborn screening, or military identification) can also reveal information about unrelated features of the genotype (e.g., risk for late-onset disease, nonpaternity) that may be both unwanted and damaging. The privacy of genetic information increasingly will become a cause for both litigation and legislation.

COMPONENTS OF THE GENETIC COUNSELING INTERACTION

Information Gathering

An integral part of genetic evaluation is, of course, the family history. This usually is recorded in the form of a pedigree so as to clarify relationships and note phenotypic

features that may be relevant to the diagnosis. Additional family history of potential genetic significance (ethnicity, consanguinity, infertility, birth defects, late-onset diseases, mental disability) should also be obtained as a matter of course. Adherence to conventions for symbols notating gender, biological relationships, pregnancy outcomes, and genotypic information, when known (Bennett et al., 1995) will ensure that any pedigree can be readily and accurately interpreted.

Medical history is routinely obtained, as is information about previous and current pregnancies, including complications and possible teratogenic or other exposures (such as smoking, radiation, or previous chemotherapy) that might have bearing on the outcome. Often, clinical features or history potentially relevant to a diagnosis must be confirmed—even before the visit—by obtaining medical records not only on the patient, but also on family members previously evaluated or treated for symptoms that may be relevant. In the context of cancer genetic counseling, it may be important to obtain not only records (such as pathology reports or test results) but actual tumor samples or slides from affected individuals.

Of equal importance to counseling success is learning about the client's or family's understanding about the reason for referral and their expectations about what will be gained through the consultation. Determining the family's beliefs about causation and assessing emotional, experiential, social, educational, and cultural issues that may affect their perception of information is a process that should be ongoing throughout the course of the evaluation.

Establishing or Verifying Diagnosis

Although a genetic diagnosis can sometimes be established or ruled out solely by reviewing medical records, evaluation usually involves at least one clinic visit. This might be for a diagnostic procedure, as in the case of prenatal testing, or for a physical examination by the clinical geneticist or another specialist experienced with the condition. Confirming a clinically suspected diagnosis often requires additional assessments, such as imaging studies, evaluations by other specialists, or examinations of particular family members. Increasingly, however, cytogenetic or molecular genetic testing alone may be sufficient not only to diagnose an affected individual or carrier, but also to provide important information about prognosis or severity. With so many of these tests commercially available, many genetic diagnoses can now be made or confirmed by the primary care physician or a specialist in a field other than genetics. The NSGC's Scope of Practice even indicates that genetic counselors may "order tests and perform clinical assessments in accordance with local, state and federal regulations." This is more likely to be appropriate in the context of prenatal diagnosis counseling or cancer risk assessment than in the general genetics clinic. Some commercial genetics laboratories have aggressively marketed genetic tests to nongeneticists and even directly to consumers—sometimes via the Internet, so testing increasingly is occurring outside of the context of genetics evaluation. This sometimes creates difficult situations in which genetic counseling must be provided post hoc to a client who was inadequately educated about testing or its implications.

Risk Assessment

In many cases, the client's concerns center not on diagnosis of an affected individual but on assessing future reproductive or personal health risk. The counselor can sometimes make such an assessment by analyzing the pedigree—taking into account the pattern of inheritance and the client's relationship to individuals with the condition. Mathematical calculations may be needed to incorporate additional information (e.g., carrier frequencies, test sensitivity and specificity, numbers of affected and unaffected individuals, the client's age) to modify the risk. Questions about carrier status may be resolved with appropriate laboratory tests. When a condition has a multifactorial basis or is genetically heterogeneous, the best risk estimates may come from epidemiologic data on other families with affected individuals. Answering concerns about potentially mutagenic or teratogenic exposures also usually relies on empirical data about the agent in question, and on evaluating the timing, duration, and dose of the exposure. In some areas of genetic counseling, such as cancer risk assessment, factors such as reproductive history, hormone use, and lifestyle issues such as smoking, obesity, or alcohol use are also important variables in risk assessment.

Information Giving

Once a diagnosis or risk is determined, the client and/or family needs to understand how it was arrived at and what the implications are for the affected person and other family members. This includes describing the condition, its variability, and its natural history—making sure that the family's prior perception of the disorder (if any) is still appropriate in light of current understanding of the genetics and treatment. It is important to make sure that, depending on the situation, the client, parents, or family are told about medical, surgical, social, and educational interventions that can correct, prevent, or alleviate symptoms. Discussions should also include available financial and social resources (e.g., support groups) to help treat and cope with the condition. When appropriate, it may be important to describe reproductive options (e.g., prenatal or preimplantation diagnosis) that could reduce risk or provide information during pregnancy. Clearly this depth of discussion would neither be warranted nor feasible in the time available for a routine prenatal session, but once a specific fetal diagnosis is made or suspected, the prospective parents should have access to as much information as they need to make a truly informed decision about their course of action.

Psychological Counseling and Support

Being given a diagnosis or learning about a personal or reproductive risk is likely to generate powerful emotional responses that must be acknowledged and dealt with if the information is to be assimilated. Part of counseling is preparing clients for these responses and helping them cope with them, often over a period of months or years. Sometimes, as in a fetal or neonatal diagnosis, critical decisions must be made rapidly

on the basis of new and distressing data. In other situations, carrier or presymptomatic testing may reveal that a person is *not* at increased risk to develop a disease or have affected children. If this new knowledge overturns long-held beliefs, it can be quite disorienting. Clients often need help in trying possible scenarios "on for size" to help them imagine how various courses of action—including just the decision to *undergo* diagnosis—may affect them and their family. The counselor must be knowledgeable about resources that can help families adjust to the reality of a condition or risk, be alert to pathological reactions that are beyond his or her skills to treat, and be able to make an effective referral when necessary.

COUNSELING CONTEXTS AND SITUATIONS

Genetic Counseling for Reproductive Issues

As genetics becomes increasingly relevant to all areas of medicine, the contexts in which genetic counseling occurs also are expanding. Genetic counselors once worked mostly in pediatrics, prenatal diagnosis, and a few specialty clinics. Today, however, people may seek counseling *before* they conceive, because of concerns about the reproductive implications of their own or their partner's family history, or to discuss carrier screening for conditions that occur more frequently in people of their ethnic background. Others may come as part of an evaluation for infertility or fetal loss, or for donor screening if they are considering using assisted reproductive techniques. With the growing use of preimplantation genetic diagnosis—not only for known genetic disorders, but also to enhance the likelihood of a successful pregnancy after in vitro fertilization—"prenatal" counseling may actually occur before conception. Also, screening pregnancies for chromosome abnormalities has shifted from the second to the first trimester and is now becoming a standard part of prenatal care for women of *any* age (Breathnach et al., 2007; Sharma et al., 2007). Indeed, discussing the host of screening options for chromosomal aneuploidy and neural tube defects has become so complicated that it is daunting for both the counselor and the pregnant couple. Despite this complexity, blood for first trimester screening is often drawn in the primary care setting in the context of "routine prenatal blood work," sometimes without the patient fully understanding the implications of screening. As a consequence, many couples who embarked on a pregnancy with no known risk factors unexpectedly find themselves in the genetic counselor's office in the first trimester discussing multiple testing options after fetal ultrasound and serum markers suggest increased risk for Down syndrome or other aneuploidies. These testing options include combining first- and second-trimester serum screening (using several possible algorithms), CVS, amniocentesis, second-trimester detailed ultrasound, and various combinations of these. Genetic counseling for prenatal diagnosis of birth defects and genetic disorders not only has become more complicated as techniques have proliferated and improved, but increasingly has shifted from large university genetics units into HMOs, private obstetricians' offices, commercial laboratories, and private hospitals.

Genetic Counseling in Pediatrics

Most genetic conditions and birth defects appear without warning. Genetic counselors have an important role to play after the birth or stillbirth of an abnormal baby or when an infant with a genetic condition dies. The counselor can help the family understand the cause of the problem (if it is known) and also help them grieve for the baby's death or the "loss" of the normal child they had anticipated. At a time when families may feel abandoned by previously trusted professionals and friends who are uncomfortable dealing with a baby's death or a birth defect, the counselor can provide not only information but also ongoing emotional support that can even continue through subsequent, usually successful, pregnancies.

Many genetic conditions are not suspected until later in childhood or even in adolescence or adulthood. In some situations, as with delayed physical or cognitive development, problems may have become evident over time. In others, a newly recognized health problem or feature of a disorder may prompt concerns about a particular diagnosis. Genetic counseling in these circumstances includes gathering information relevant to establishing the diagnosis, anticipating its impact on the patient or family, addressing their fears and distress, educating them about the condition and its implications, and ensuring that they access necessary medical and social services. Because genetic counselors understand the unique genetic, psychosocial, and medical issues that attend many chronic conditions, they are often part of teams of professionals who provide ongoing management for diseases such as cystic fibrosis, craniofacial or bleeding disorders, muscular dystrophies, inherited metabolic conditions, and hemoglobinopathies.

Genetic Counseling for Adult-Onset Diseases

A newer arena for genetic counseling is in genetic testing for conditions that develop later in life. As molecular tests have become available for disorders such as Huntington disease, familial amyotrophic lateral sclerosis, and numerous hereditary cancer predispositions, healthy individuals who are at risk may consider learning about their genotype so as to diminish anxiety, remove uncertainty, or make personal and medical decisions. Numerous complex genetic and psychosocial issues arise in helping families consider testing and cope with the results. Many physicians who traditionally have cared for affected individuals in these families feel ill equipped to provide the necessary education and counseling that should surround testing. Consequently, genetic counselors now find themselves working in settings such as cancer centers, dialysis units, and adult neurology, cardiology, or dermatology clinics that historically may not have had close relationships with genetics. Many clinics are now hiring genetic counselors directly, rather than "borrowing" them from genetics units, with the result that some counselors now work more closely with an oncologist, neurologist, or cardiologist than they do with a clinical geneticist.

These new work settings are interesting in that genetic counselors are removed from the traditional "genetics team" and may be looked to for diagnostic expertise that formerly was provided by clinical geneticists. Up until recently, diagnosing

most genetic conditions required the skill in physical diagnosis and acumen in synthesizing complex historical and laboratory information that physicians have. For many genetic conditions, this undoubtedly will always be the case. While genetic counselors are trained to understand genetic test results, their training does not develop these other diagnostic skills. However, with diagnosis of some disease predispositions, carrier states, and adult-onset genetic disorders able to be established solely through molecular or other types of genetic tests, genetic counselors in certain clinical settings are able to function more autonomously. The NSGC has indicated that ordering diagnostic tests and other clinical assessments, "in accordance with local, state and federal regulations," is within the genetic counselor's scope of practice.

PROVIDERS OF GENETIC COUNSELING

Geneticists

Elements of genetic counseling—risk assessment, information about genetic disorders and reproductive options, treatment for psychological distress related to these issues—are provided by many types of health care workers in diverse settings. However, with genetics now recognized as a distinct medical specialty, people with a genetic condition or birth defect ideally should be seen at some point by one or more professionals with specialized training in genetic diagnosis and counseling. In many centers, a genetics *team* that includes many of the geneticists described below, each with distinct disciplinary backgrounds, roles, and areas of expertise, provides these services. The NSGC Code of Ethics has a section relating to our obligations in regard to relationships with these colleagues. Specifically mentioned is the importance of respecting and valuing their knowledge, perspectives, contributions, and areas of competence and collaborating with them to provide the highest quality of service.

Genetic Counselors The first graduate program to educate master's-level professionals in human genetics and genetic counseling was started at Sarah Lawrence College in 1969 (Marks and Richter, 1976). There are now over 50 such programs in the United States, Canada, Cuba, the United Kingdom, the Netherlands, Norway, Sweden, France, Spain, Israel, Saudi Arabia, South Africa, Australia, Japan, China, and Taiwan. Most programs outside of North America have started since the beginning of the millennium, and several more countries are now actively planning to train counselors.

According to the NSGC Professional Status Survey (2006), to which about two-thirds of members responded, 79% of genetic counselors provide direct clinical services. Most work in an academic medical center (38%), but collectively even more work in a private (20%) or public (11%) hospital/medical facility, a diagnostic laboratory (8%), a private physician's office (5%), or an HMO (3%). About 55% are responsible for teaching or providing clinical supervision, and roughly one in five (usually those associated with an academic medical center) has some type of faculty

appointment. Over a quarter coordinate clinics and/or research studies. Other primary roles include healthcare administration, public policy, management, and client services or marketing for a commercial laboratory. Some counselors see patients in just one specialty area, while others cover several areas. The most frequently reported was prenatal diagnosis (54%), followed by cancer genetics (39%), pediatrics (34%), adult genetics (24%), and specialty clinics (13%). Less frequent activities (reported in each case by less than 10% of counselors) were public health and screening programs and counseling related to infertility, neurogenetics, cardiology, psychiatric genetics, or possible teratogenic exposures.

Clinical Geneticists Physicians who have completed accredited residency and/or fellowship programs in North America may become eligible for certification in clinical genetics by the American Board of Medical Genetics (ABMG) or the Canadian College of Medical Genetics (CCMG). In the past, many of these physicians first trained in pediatrics, internal medicine, obstetrics, or another specialty before entering genetics. Recognition of the ABMG by the American Board of Medical Specialties in the early 1990s meant that residencies could have clinical genetics as the *primary* specialty. Some institutions also offer one or more combined residencies with both genetics and another specialty as the focus. There are several such programs in the U.S. and also in Canada (where clinical genetics training is under the aegis of the CCMG and the Royal College of Physicians).

Board certification in clinical genetics requires the physician to have knowledge and experience in diagnosing and treating genetic conditions and birth defects, as well as a thorough understanding of the underlying genetics principles. Genetic counseling is assumed to be part of their fellowship training. Clinical geneticists often have particular areas of interest, such as dysmorphology, neurogenetics, metabolic or adult disorders, or prenatal diagnosis, but should also be able to provide expertise on diagnosis and management of a wide range of genetic conditions.

Other Genetics Subspecialists In addition to clinical geneticists (who must be physicians), the ABMG certifies several other categories of genetics professionals. These include cytogeneticists, molecular geneticists, and biochemical geneticists—many of whom direct genetics diagnostic laboratories. People usually seek ABMG certification if they intend to be involved in clinical activities—either seeing patients or doing diagnostic testing—so even laboratory-oriented certification examinations assess knowledge of genetic counseling in addition to expertise in the appropriate subspecialty(ies). Historically, the ABMG has been unusual among medical specialty boards in certifying Ph.D.s as well as M.D.s and in having been at one time (1982 to 1990) the certifying board for genetic counselors. Some geneticists who are certified in laboratory subspecialties also counsel and treat patients with diseases they diagnose. By the same token, clinical geneticists who perform and interpret diagnostic tests or who specialize in diagnosing and treating metabolic or chromosomal disorders may have additional certification(s) in these subspecialties, even if they are not directly involved in a diagnostic laboratory. Some laboratories doing genetic

testing may be directed or staffed by clinical pathologists who have acquired knowledge and skills in molecular genetics or cytogenetics.

Genetics Nurses There are enough nurses working in genetics to have their own professional society (The International Society of Nurses in Genetics, or ISONG), although relatively few are certified in *genetic counseling*. This is because eligibility for both the ABMG and the American Board of Genetic Counseling (ABGC) has required master's-level training in genetics, usually from an accredited genetic counseling program. However, advanced practice and other specialty nurses often work in clinics and programs where genetic disorders and birth defects are diagnosed and treated. Many have acquired their knowledge of genetics through years of clinical experience, and a few actually hold a graduate degree in genetics nursing from one of the handful of programs that have provided such training. Nurses' additional skills in physical and psychosocial assessment, case management, patient education, clinic administration, and community health are highly valued in specialty and outreach clinics, and in genetics screening programs. Those with specialization in areas such as infant special care, oncology, or midwifery may be astute "case-finders" of patients in need of genetics services and helpful allies in their care.

The Genetics Nursing Credentialing Commission uses a portfolio-based mechanism for appropriately prepared nurses to become credentialed in genetic nursing. Those with a graduate degree from an accredited program and 300 hours of training in a practice at least half devoted to genetics can qualify for a credential as an Advanced Practice Nurse in Genetics (APNG) by providing a logbook of 50 genetics cases and an in-depth written description of four cases, and by documenting sufficient recent genetics coursework or continuing education. Nurses with only a bachelor of nursing degree can qualify as a Genetics Clinical Nurse (GCN) through a similar process. This portfolio-based approach to credentialing is similar to what is used for genetic counselors in the UK and some other countries that do not have examination-based certification.

NonGeneticists

Many patients receive "genetic counseling" in the context of primary or specialty care from health providers who are not geneticists. Examples of such "genetics services" would include molecular or cytogenetic diagnostic testing, screening for potential genetic risks via a family history questionnaire, interview or blood test (e.g., to look for hemoglobinopathies or to measure maternal serum markers during pregnancy), or advising patients about reproductive risks and screening or testing options (as is now done in many prenatal care settings). Commercial availability of a host of tests for genetic diseases or predispositions and their use by nongeneticists means that people more frequently will be asked to consider these tests or will be given results by health providers who have had relatively little training in medical genetics. Many of these providers have not been exposed to the idea of nondirective counseling, and they frequently work under pressures that limit the time that can be spent in discussion. Economic constraints of managed

care, fear of litigation, and patient demand are factors that may encourage providers to try to provide genetic tests themselves. For these reasons, geneticists and genetic counselors have an obligation to help educate other health care providers in (1) recognizing potential genetic risks; (2) being aware of phenomena such as variability, heterogeneity, and penetrance that can complicate genetic counseling; (3) understanding the complexities and limitations as well as the benefits of genetic testing; (4) appreciating the philosophy of nondirective counseling; (5) being sensitive to inherent ethical dilemmas; and (6) knowing when they should refer a client to a geneticist or genetic counselor.

The NSGC Code of Ethics specifically refers to genetic counselors' duty to share their knowledge of genetics with other health care providers and to provide mentorship and guidance with regard to genetics health care. The National Coalition for Health Professional Education in Genetics (NCHPEG) has developed useful resources that provide guidance for developing the genetics content of curricula and continuing education programs for *all* health care providers ("Core Competencies in Genetics Essential for All Health Care Professionals" and "Core Principles of Genetics for Health Professionals"). The NCHPEG website is a very useful source of materials that genetic counselors can access to help them educate nongeneticist colleagues.

PROFESSIONAL AND EDUCATIONAL LANDMARKS IN GENETIC COUNSELING

Development of Training Programs and Curricula for Genetic Counseling

In 1971, the year in which the first 10 "genetic associates" graduated from Sarah Lawrence, a report of the National Institute of General Medical Services predicted that by 1988, 68% more geneticists would be needed to provide appropriate services. By 1973, four more genetic counseling programs had been started. The next year, a meeting of various faculty, students, and graduates from four of the five existing programs was held at the California state conference grounds at Asilomar to discuss training goals and expectations for this new professional. Representatives from state and federal health care agencies, genetics centers, volunteer health organizations and legislators, as well as counseling program directors and graduates attended a second Asilomar meeting, sponsored by the March of Dimes in 1976. Both these meetings emphasized the importance of planning and program evaluation for genetic counseling training.

In the spring of 1979, the Office of Maternal and Child Health (MCH) sponsored a much more comprehensive meeting, involving about 50 people, in Williamsburg, Virginia. Participants represented constituencies similar to those in 1976, but also included planners from health maintenance organizations and the health insurance industry, nurses and social workers who provided genetics services, and representatives of the NSGC, which had been formed just the year before. Four panels were

assigned different tasks: (1) evaluating the curricula of existing programs in light of graduates' experience; (2) exploring how genetic counselors' services could be reimbursed; (3) recommending ways of ensuring continuing education for genetic counselors; and (4) suggesting a means of evaluating the quality of programs and the competence of their graduates (Dumars et al., 1979). Of all the recommendations to come from the Williamsburg meeting, the guidelines that were established for the curricular content and structure of genetic counseling training had the most lasting impact. The Williamsburg curricular guidelines influenced planning for the many new training programs that started over the next decade. In 1989 yet another conference was held in Asilomar—this time under sponsorship of the NSGC as well as MCH. The purpose of this meeting was to reevaluate the Williamsburg recommendations for program curricula and clinical training and to explore innovative ways for addressing newer aspects of genetics practice, such as cross-cultural counseling and molecular genetics. Additional issues that were discussed included the pros and cons of instituting a doctoral degree in genetic counseling, and potential solutions for shortfalls in genetics "person-power"—including the possibility of more limited training for "genetics assistants" or "aides" who would assume routine tasks and perhaps bring more diverse cultural perspectives to counseling (Walker et al., 1990).

The National Society of Genetic Counselors

A milestone in the evolution of any profession is the formation of its own society. For genetic counselors, this came in 1979 when the NSGC was incorporated. The goals of the new society were "to further the professional interests of genetic counselors, to promote a network of communication within the genetic counseling profession and to deal with issues related to human genetics" (Heimler, 1979).[3] Over the years, the NSGC has, in fact, done this. In 1980, the newly formed NSGC—then numbering only about 200—lobbied successfully for genetic counselors to be included among subspecialties that would be certified by the ABMG. The NSGC has helped achieve representation by genetic counselors on the Boards of Directors of the American Society of Human Genetics (ASHG) and on numerous committees of the ASHG, the American College of Medical Genetics (ACMG), and various government advisory boards. The NSGC sponsors an annual meeting to provide continuing education for its members and a forum for discussing research and clinical issues of interest. Since 1992, it has published its own journal that is indexed in at least ten databases, including PubMed. By 1991, the NSGC had developed a code of ethics for the profession. Most importantly, the Society has become recognized as the voice of the profession and a resource for information about genetic counseling issues by the media, the public, and other health, public policy, and genetics professionals.

[3] Audrey Heimler provides a wonderful account of the early days of the NSGC and its contribution to the evolution of the genetic counseling profession in the *Journal of Genetic Counseling* (1997, 6:315–336).

Accreditation of Genetic Counseling Training Programs

In 1993, the ABGC was incorporated with the goal of certifying genetic counselors; this had been the province of the ABMG since 1980. This change was necessary so that the ABMG could be eligible for recognition by the American Board of Medical Specialties, which does not allow member boards to certify non-doctoral-level professionals, but it required a majority vote of all ABMG diplomates, including counselors. The vote passed in 1992, following a year of rancorous debate, and the ABGC was incorporated in 1993. However painful, forming the ABGC was an important landmark in the evolution of the genetic counseling profession because it not only allowed for more autonomous decision-making about certifying genetic counselors, but also provided an opportunity to accredit genetic counseling *programs*. The ABMG accredits fellowships (and now residencies) for Ph.D. and M.D. geneticists, but these are not degree-granting programs. In regard to *genetic counselors'* training, the ABMG had chosen to limit its oversight to approving sites in which genetic counseling students got clinical experience and to graduates' case logbooks that they submitted to establish board eligibility. It did not review or accredit genetic counseling training programs, which were proliferating rapidly in the early 1990s. Given that potential programs were seeking guidance about program design and that potential students wanted reassurance about newer programs they might be considering, the ABGC felt that it should undertake the task of program accreditation.

The ABGC's newly formed accreditation committee explored with other boards that accredited allied health programs and with outside consultants how the ABGC might approach accreditation. From these investigations it became clear that flexibility, variety, and innovation in training were more likely to occur if the accrediting body were to evaluate a program's ability to develop various *professional competencies* in its graduates rather than simply determining how well it adhered to prescribed guidelines for curriculum and clinical experiences. However, no expectations for "entry-level" competencies in genetic counseling had ever been clearly defined. To this end, the ABGC sponsored a 1994 meeting that included directors of all existing genetic counseling programs, the ABGC Board, and consultants from outside the genetic counseling field who had expertise in clinical supervision and accreditation. The goal was to develop consensus about what new graduates should be able to do. By analyzing the counselor's role in various clinical scenarios, participants identified areas of required knowledge and skills (Fiddler et al., 1996), and from these analyses 27 "competencies" were described. Competencies were further refined by the ABGC (Fine et al., 1996) to form the basis of a document that would be used to guide nascent programs and those seeking accreditation (American Board of Genetic Counseling, 1996). The ongoing validity of these was affirmed at another small retreat sponsored by the ABGC Board in July 2005, when the competencies were revisited and thought to need minimal, if any revision. Participants observed that while the *knowledge base* for trainees has changed, expectations for *basic skills* remain the same. Since helping to develop these competencies is what this book is all about, their description is appended to this chapter. They are also posted on the ABGC's website.

Worldwide Genetic Counseling Training

The need for and potential in having professionally trained genetic counselors as part of the workforce delivering genetics services is now recognized in many different nations. Since 1995 at least 26 new programs have been started in 17 countries outside of North America, with genetic counselors now being trained on five continents (Transnational Alliance for Genetic Counselling, 2009)! Directors of many of these programs met as a group for the first time in Manchester, England in May 2006 to learn about each other's curricula and experiential training, genetics service delivery models, and mechanisms for genetic counselor credentialing. A transnational alliance of genetic counseling (TAGC) was born at this meeting, and one outcome is that information about training programs around the world can now be found on the TAGC website.

Not all countries with training programs award a master's-level degree; in some cases genetic counseling education results in a diploma or certificate that documents appropriate knowledge, clinical experiences, and counseling skills. During the 1980s and 1990s, Regina Kenen studied the emergence and evolution of the genetic counseling profession, both in the United States and in Australia, finding that it involved three identifiable stages (Kenen, 1986, 1997). She described an initial "emergent" phase with professional issues and rivalries about who should provide genetic counseling—their relative status, power, and requisite skills. This was followed by a "consolidation" phase in which the service delivery model was refined and professional roles clarified. The final "institutionalized" stage was one in which the model and profession had become established so that goals and standards were understood by all and there was minimal conflict between the various constituencies.[4] Discussions in Manchester found several countries still in the emergent stage, but also emphasized that even in countries such as the U.S. where training programs have existed for nearly four decades, it was still necessary to adjust to changes in health care delivery, advances in genetics, and, increasingly, excursions of genetic counselors into "nontraditional" roles in research, public health, policy, and areas of medicine in which genetics historically has not been prominent.

By the time of the first meeting in 2006, seven countries had developed a mechanism to identify who was qualified to provide genetic counseling. While the specific names for these mechanisms vary, the purpose is the same: to ensure a minimum standard of safe practice so as to protect the public, and to elevate and promote awareness of the profession. Different countries have different pathways to establish eligibility for recognition of professional competence. In the U.S., Australasia, and South Africa, the only route is via a master's degree or graduate diploma in genetic counseling. Other countries (e.g., the U.K., Canada, the Netherlands, and Japan) have a second potential pathway allowing those with a bachelor's degree (in nursing or another relevant field) AND postqualification clinical experience, and in some cases counseling

[4] Margaret A. Sahhar provides an insightful overview of the development of genetic counseling training in Australia—comparing their education and training requirements to those in the U.K., Canada, and the U.S., in the *Journal of Genetic Counseling* (2005, 14:283–294).

training, a route to credentialing.[5] In general, these alternate pathways are eventually phased out, as they were in the U.S., as an increasing proportion of genetic counselors have had formal academic training and earned a post-bachelor's degree in the field.

Professional Recognition of Competence in Genetic Counseling

ABGC Certification The ABGC has certified genetic counselors in the U.S. and Canada since 1993. Achieving certification requires that a candidate's application be approved by the ABGC Credentials Committee (credentialing) to establish "active candidate status". Once approved, the candidate must pass a written examination. Starting in 2009, this will be a single, comprehensive, four-hour examination, developed and administered by the ABGC (American Board of Genetic Counseling 2009). Prior to 2009, certification required the candidate to pass both a general genetics examination–developed and administered by the ABMG, and a genetic counseling examination, created by the ABGC.

When the ABMG first offered certification in 1981, people doing genetic counseling came from a variety of training backgrounds. Many had graduated from genetic counseling programs, but others were nurses, social workers, or simply had a post-baccalaureate degree in genetics. The ABMG established that eligibility for certification in genetic counseling required at least a master's degree in a relevant discipline, provision of genetic counseling in 50 diverse cases documented in a logbook submitted with the application, and letters of reference from three other geneticists.

Since then, requirements for certification have become more stringent, with the ABGC now requiring applicants to have graduated from a genetic counseling training program that was accredited when they entered training. Through the 2009 examination cycle, logbook cases must have been acquired in clinical sites approved by the ABGC, either via accreditation of the training program(s) with which the sites are affiliated, or by Board review of an application submitted for an "ad hoc" site utilized by one particular trainee over a specified period. The logbook has required the nature of the candidate's involvement in each case to be clearly described, with cases demonstrating a participation in a variety of counseling roles and clinical situations. Cases must be supervised by an ABGC or ABMG certified individual. Starting with the 2010 examination, the ABGC will no longer require candidates to submit their logbooks. It will be incumbent on training programs to ensure that their training sites are approved and that their trainees have appropriate clinical supervision and obtain the necessary breadth and depth of cases. The ABGC publishes a bulletin describing current requirements and providing application forms and instructions for the examination, which will be given annually starting in 2010. The ABGC website should be relied on for up-to-date information.

[5] This information comes from a Manchester meeting workshop organized by Lauren Kerzin-Storrar and Anna Middleton to explore credentialing models and consider the possibility of transnational reciprocity for counselors credentialed in one country who wish to practice in another. Ms. Kerzin-Storrar kindly provided her draft summary of the proceedings to the author, who also participated in the workshop.

With the advent of more international training programs, increasing numbers of counselors who had trained and practiced outside North America expressed a desire to be qualified to practice in the U.S. or Canada. After receiving many requests for access to ABGC certification, the Board developed a mechanism for appropriately trained individuals to apply for 'International Genetic Counselor Certification' (IGCC). This mechanism was also available to graduates of a few Canadian programs that were not ABGC-accredited at the time the trainee matriculated. Eligibility criteria were strict; the person needed to have received at least a master's degree from a formal genetic counseling program outside the U.S., and must also have acquired a 50 case logbook under appropriate clinical supervision in an ABGC-accredited genetic counseling program over no less than a six-month period. For a variety of reasons, the ABGC discontinued the IGCC program in September 2008. The Canadian Association of Genetic Counselors (CAGC) and the Genetic Counseling Registration Board in the U.K. have, or may soon have, mechanisms for credentialing counselors trained outside their respective countries.

Licensure and Registration In the U.S., these two terms refer to establishment of an individual's credentials and formal recognition as a member of a profession by a state governmental body, such as a licensure board, Department of Consumer Affairs, etc. Such agencies are charged with protecting the public by ensuring that specified professionals provide a minimum standard of care within their scope of practice. Hospitals and other health care organizations generally expect that providers who seek to work in their institutions will be licensed or registered, as do many "third-party payers" such as insurance companies and publicly funded medical programs. Usually, it is the state's legislature that decides whether a group needs to be licensed to protect the interests of the citizens it represents. The legislature's motivation to pass a law mandating licensure is influenced by *public demand* (both on the part of the group seeking to be licensed and the public at large), by the *potential for harm* from unqualified practitioners, by the *number of practitioners* in the state, and also by having *a committed legislator* willing to carry the bill. Passage of a law is usually only the first step; regulations must then be written to define who will be licensed and how, and a board must be established to review credentials and investigate complaints. In 2000, California became the first state to pass legislation for licensure of genetic counselors. The first state to actually *issue* genetic counseling licenses was Utah, whose law provides 'title protection' that bars an unlicensed individual from calling him- or herself a "genetic counselor" and requires genetic counselors to be licensed in order to practice. As of March 2009, genetic counselors in five additional states have been able to obtain licenses (NSGC 2009). A few other states are still developing regulations to *implement* a licensure law that has been passed, and several more have licensure bills introduced or pending.

Other Forms of Credentialing The United Kingdom and South Africa use the term "registration" in the same way as "certification" is used in the United States and Canada, although registration may be required in order to practice. Some countries (e.g., U.S., Canada, Japan) require examination by a certifying board, while others

(e.g., Australia, U.K.) require postqualification work experience and portfolios detailing clinical training in approved clinical genetics centers (Sahhar et al., 2005). In the U.S. and South Africa, nurses working in genetics have their own separate process for credentialing, although some U.S. nurses with a master's degree were certified in genetic counseling by the ABMG in the 1980s.

PROFESSIONAL GROWTH AND SKILL ACQUISITION

The entry-level genetic counseling competencies that were defined at the 1996 ABGC consensus development conference are used not only to guide and evaluate training programs but also to monitor the progress of students as they acquire the knowledge and skills they will need to be effective genetic counselors. Training programs use the competencies in planning the timing and content of classes and clinical experiences so that students will have been exposed to relevant coursework by the time that they need to use this knowledge in interacting with patients. The competencies also frequently form the basis of evaluation forms used in clinical rotations. They are more than just a checklist of skills and reflect a need for the student to be able to call upon and integrate knowledge about genetic mechanisms, inheritance patterns, disease manifestations, family dynamics, and coping mechanisms with skills in obtaining and interpreting histories, pedigree construction, risk assessment, interviewing, psychosocial evaluation, explaining technical information, etc., in order to manage a counseling session. Other competencies address areas such as the ability to find and synthesize information, identify community resources and advocate for clients, function as part of a team, manage and document a case, provide public and professional education, evaluate and participate in research, show cultural awareness, and behave according to the philosophical, ethical, and legal tenets of the profession.

The last two competencies have to do with recognizing the limits of one's own expertise and taking responsibility for life-long learning. While genetic counselors enter the field with an impressive armamentarium of skills and knowledge, there is no way that two years of training can prepare them for all counseling situations or for future developments in genetics that cannot even be imagined today. Ongoing self-education is critical, and counselors must stay abreast of the literature, routinely attend professional meetings, and communicate with genetics colleagues in order to provide quality service. The NSGC has special interest groups (SIGs) for many subspecialty areas. Many of these SIGs have active e-mail list-serves that provide invaluable information and a forum to learn, ask questions, and discuss issues of common interest. A number of the SIGs sponsor workshops at the NSGC Annual Education Meeting to update their members and other counselors on recent advances and changes in counseling practice. There are also other avenues to explore and develop competence in a rapidly expanding variety of related areas. Some counselors elect to obtain additional formal training to enhance their ability to do psychotherapy, research, or administration, or perhaps to enable them to function in a new domain, such as behavioral or clinical research or commercial genetics. Maintaining membership in professional societies and being active on their committees afford opportunities to work

with colleagues from around the country and to develop leadership skills. Involvement in education, advocacy, and political activism can also bring personal rewards and lead to recognition in the community and beyond.

As members of a relatively small profession that deals with issues at the cutting edge of science, medicine, and ethics we are all required to continue to grow and to take responsibility for helping other health professionals, policy makers, and clients understand genetics and its implications. The challenges are many, but the personal and professional rewards are enormous.

APPENDIX

Practice-Based Competencies (ABGC, 1996)

"An entry-level genetic counselor must demonstrate the practice-based competencies listed below to manage a genetic counseling case before, during, and after the clinic visit or session. Therefore, the didactic and clinical training components of a curriculum must support the development of competencies that are categorized into the following domains: Communication Skills; Critical-Thinking Skills; Interpersonal, Counseling, and Psychosocial Assessment Skills; and Professional Ethics and Values. Some competencies may pertain to more than one domain. These domains represent practice areas that define activities of a genetic counselor. The italicized facet below each competency elaborates on skills necessary for achievement of each competency. These elaborations should assist program faculty in curriculum planning, development, and program and student evaluation.

Domain I: Communication Skills

a. Can establish a mutually agreed upon genetic counseling agenda with the client.

   The student is able to contract with a client or family throughout the relationship; explain the genetic counseling process; elicit expectations, perceptions and knowledge; and establish rapport through verbal and non-verbal interaction.

b. Can elicit an appropriate and inclusive family history.

   The student is able to construct a complete pedigree; demonstrate proficiency in the use of pedigree symbols, standard notation, and nomenclature; structure questioning for the individual case and probable diagnosis; use interviewing skills; facilitate recall for symptoms and pertinent history by pursuing a relevant path of inquiry; and in the course of this interaction, identify family dynamics, emotional responses, and other relevant information.

c. Can elicit pertinent medical information including pregnancy, developmental, and medical histories.

   The student is able to apply knowledge of the inheritance patterns, etiology, clinical features, and natural history of a variety of genetic disorders, birth

defects, and other conditions; obtain appropriate medical histories; identify essential medical records and secure releases of medical information.

d. Can elicit a social and psychosocial history.

The student is able to conduct a client or family interview that demonstrates an appreciation of family systems theory and dynamics. The student is able to listen effectively, identify potential strengths and weaknesses, and assess individual and family support systems and coping mechanisms.

e. Can convey genetic, medical, and technical information including, but not limited to, diagnosis, etiology, natural history, prognosis, and treatment/management of genetic conditions and/or birth defects to clients with a variety of educational, socioeconomic, and ethnocultural backgrounds.

The student is able to demonstrate knowledge of clinical genetics and relevant medical topics by effectively communicating this information in a given session.

f. Can explain the technical and medical aspects of diagnostic and screening methods and reproductive options including associated risks, benefits, and limitations.

The student is able to demonstrate knowledge of diagnostic and screening procedures and clearly communicate relevant information to clients. The student is able to facilitate the informed-consent process. The student is able to determine client comprehension and adjust counseling accordingly.

g. Can understand, listen, communicate, and manage a genetic counseling case in a culturally responsive manner.

The student can care for clients using cultural self-awareness and familiarity with a variety of ethnocultural issues, traditions, health beliefs, attitudes, lifestyles, and values.

h. Can document and present case information clearly and concisely, both orally and in writing, as appropriate to the audience.

The student can present succinct and precise case-summary information to colleagues and other professionals. The student can write at an appropriate level for clients and professionals and produce written documentation within a reasonable time frame. The student can demonstrate respect for privacy and confidentiality of medical information.

i. Can plan, organize, and conduct public and professional education programs on human genetics, patient care, and genetic counseling issues.

The student is able to identify educational needs and design programs for specific audiences, demonstrate public speaking skills, use visual aids, and identify and access supplemental educational materials.

Domain II: Critical-Thinking Skills

a. Can assess and calculate genetic and teratogenic risks.

The student is able to calculate risks based on pedigree analysis and knowledge of inheritance patterns, genetic epidemiologic data, and quantitative genetics principles.

b. Can evaluate a social and psychosocial history.

The student demonstrates understanding of family and interpersonal dynamics and can recognize the impact of emotions on cognition and retention, as well as the need for intervention and referral.

c. Can identify, synthesize, organize and summarize pertinent medical and genetic information for use in genetic counseling.

The student is able to use a variety of sources of information including client/family member(s), laboratory results, medical records, medical and genetic literature and computerized databases. The student is able to analyze and interpret information that provides the basis for differential diagnosis, risk assessment and genetic testing. The student is able to apply knowledge of the natural history and characteristics/symptoms of common genetic conditions.

d. Can demonstrate successful case management skills.

The student is able to analyze and interpret medical, genetic and family data; to design, conduct, and periodically assess the case management plan; arrange for testing; and follow up with the client, laboratory, and other professionals. The student should demonstrate understanding of legal and ethical issues related to privacy and confidentiality in communications about clients.

e. Can assess client understanding and response to information and its implications to modify a counseling session as needed.

The student is able to respond to verbal and nonverbal cues and to structure and modify information presented to maximize comprehension by clients.

f. Can identify and access local, regional, and national resources and services.

The student is familiar with local, regional, and national support groups and other resources, and can access and make referrals to other professionals and agencies.

g. Can identify and access information resources pertinent to clinical genetics and counseling.

The student is able to demonstrate familiarity with the genetic, medical and social-science literature, and on-line databases. The student is able to review the literature and synthesize the information for a case in a critical and meaningful way.

Domain III: Interpersonal, Counseling, and Psychosocial Assessment Skills

a. Can establish rapport, identify major concerns, and respond to emerging issues of a client or family.

The student is able to display empathic listening and interviewing skills, and address clients' concerns.

b. Can elicit and interpret individual and family experiences, behaviors, emotions, perceptions, and attitudes that clarify beliefs and values.

The student is able to assess and interpret verbal and non-verbal cues and use this information in the genetic counseling session. The student is

able to engage clients in an exploration of their responses to risks and options.

c. Can use a range of interviewing techniques.
   The student is able to identify and select from a variety of communication approaches throughout a counseling session.

d. Can provide short-term, client-centered counseling and psychological support.
   The student is able to assess clients' psychosocial needs and recognize psychopathology. The student can demonstrate knowledge of psychological defenses, family dynamics, family theory, crisis-intervention techniques, coping models, the grief process, and reactions to illness. The student can use open-ended questions; listen empathically; employ crisis-intervention skills; and provide anticipatory guidance.

e. Can promote client decision-making in an unbiased, non-coercive manner.
   The student understands the philosophy of non-directiveness and is able to recognize his or her values and biases as they relate to genetic counseling issues. The student is able to recognize and respond to dynamics, such as countertransference, that may affect the counseling interaction.

f. Can establish and maintain inter- and intradisciplinary professional relationships to function as part of a health-care delivery team.
   The student behaves professionally and understands the roles of other professionals with whom he or she interacts.

Domain IV: Professional Ethics and Values

a. Can act in accordance with the ethical, legal, and philosophical principles and values of the profession.
   The student is able to recognize and respond to ethical and moral dilemmas arising in practice and seek assistance from experts in these areas. The student is able to identify factors that promote or hinder client autonomy. The student demonstrates an appreciation of the issues surrounding privacy, informed consent, confidentiality, real or potential discrimination, and other ethical/legal matters related to the exchange of genetic information.

b. Can serve as an advocate for clients.
   The student can understand clients' needs and perceptions and represent their interests in accessing services and responses from the medical and social service systems.

c. Can introduce research options and issues to clients and families.
   The student is able to critique and evaluate the risks, benefits, and limitations of client participation in research; access information on new research studies; present this information clearly and completely to clients; and promote an informed-consent process.

d. Can recognize his or her own limitations in knowledge and capabilities regarding medical, psychosocial, and ethnocultural issues and seek consultation or refer clients when needed.

The student demonstrates the ability to self-assess and to be self-critical. The student demonstrates the ability to respond to performance critique and integrates supervision feedback into his or her subsequent performance. The student is able to identify and obtain appropriate consultative assistance for self and clients.

e. Can demonstrate initiative for continued professional growth.

The student displays a knowledge of current standards of practice and shows independent knowledge-seeking behavior and lifelong learning."

Genetic Counselors' Scope of Practice (NSGC, 2007)

"This 'Genetic Counselors' Scope of Practice' statement outlines the responsibilities of individuals engaged in the practice of genetic counseling. Genetic counselors are health professionals with specialized education, training and experience in medical genetics and counseling who help people understand and adapt to the implications of genetic contributions to disease.[6] Genetic counselors interact with clients and other healthcare professionals in a variety of clinical and non-clinical settings, including, but not limited to, university-based medical centers, private hospitals, private practice, and industry settings. The instruction in clinical genetics, counseling, and communication skills required to carry out the professional responsibilities described in this statement is provided in graduate training programs accredited by the American Board of Genetic Counseling (ABGC)[7] or the equivalent, as well as through professional experience and continuing education courses.

The responsibilities of a genetic counselor are threefold: (i) to provide expertise in clinical genetics; (ii) to counsel and communicate with patients on matters of clinical genetics; and (iii) to provide genetic counseling services in accordance with professional ethics and values. Specifically:

Section I: Clinical Genetics

1. Explain the nature of genetics evaluation to clients. Obtain and review medical and family histories, based on the referral indication, and document the family history using standard pedigree nomenclature.
2. Identify additional client and family medical information relevant to risk assessment and consideration of differential diagnoses, and assist in obtaining such information.
3. Research and summarize pertinent data from the published literature, databases, and other professional resources, as necessary for each client.
4. Synthesize client and family medical information and data obtained from additional research as the basis for risk assessment, differential diagnosis, genetic testing options, reproductive options, follow-up recommendations, and case management.

[6] NSGC Definition of Genetic Counseling. *Journal of Genetic Counseling* April 2006; 77–82.
[7] American Board of Genetic Counseling website.

5. Assess the risk of occurrence or recurrence of a genetic condition or birth defect, using a variety of techniques, including knowledge of inheritance patterns, epidemiologic data, quantitative genetics principles, statistical models, and evaluation of clinical information, as applicable.
6. Explain to clients, verbally and/or in writing, medical information regarding the diagnosis or potential occurrence of a genetic condition or birth defect, including etiology, natural history, inheritance, disease management and potential treatment options.
7. Discuss available options and delineate the risks, benefits and limitations of appropriate tests and clinical assessments. Order tests and perform clinical assessments in accordance with local, state and federal regulations.
8. Document case information clearly and concisely in the medical record and in correspondence to referring physicians, and discuss case information with other members of the healthcare team, as necessary.
9. Assist clients in evaluating the risks, benefits and limitations of participation in research, and facilitate the informed consent process.
10. Identify and access local, regional, and national resources such as support groups and ancillary services; discuss the availability of such resources with clients; and provide referrals, as necessary.
11. Plan, organize and conduct public and professional education programs on medical genetics, patient care and genetic counseling issues.

Section II: Counseling and Communication

1. Develop a genetic counseling agenda with the client or clients that includes identification and negotiation of client/counselor priorities and expectations.
2. Identify individual client and family experiences, behaviors, emotions, perceptions, values, and cultural and religious beliefs in order to facilitate individualized decision making and coping.
3. Assess client understanding and response to medical information and its implications, and educate client appropriately.
4. Utilize appropriate interviewing techniques and empathic listening to establish rapport, identify major concerns and engage clients in an exploration of their responses to the implications of the findings, genetic risks, and available options/interventions.
5. Identify the client's psychological needs, stressors and sources of emotional and psychological support in order to determine appropriate interventions and/or referrals.
6. Promote client-specific decision making in an unbiased non-coercive manner that respects the client's culture, language, traditions, lifestyle, religious beliefs and values.
7. Use knowledge of psychological structure to apply client-centered techniques and family systems theory to facilitate adjustment to the occurrence or risk of occurrence of a congenital or genetic disorder.

Section III: Professional Ethics and Values

1. Recognize and respond to ethical and moral dilemmas arising in practice, identify factors that promote or hinder client autonomy, and understand issues surrounding privacy, informed consent, confidentiality, real or potential discrimination and potential conflicts of interest.
2. Advocate for clients, which includes understanding client needs and perceptions, representing their interests in accessing services, and eliciting responses from the medical and social service systems as well as the community at large.
3. Recognize personal limitations in knowledge and/or capabilities and seek consultation or appropriately refer clients to other providers.
4. Maintain professional growth, which includes acquiring relevant information required for a given situation, keeping abreast of current standards of practice as well as societal developments, and seeking out or establishing mechanisms for peer support.
5. Respect a client's right to confidentiality, being mindful of local, state and federal regulations governing release of personal health information.

This Scope of Practice statement was approved in June 2007 by the National Society of Genetic Counselors (NSGC)—the leading voice, advocate and authority for the genetic counseling profession. It is not intended to replace the judgment of an individual genetic counselor with respect to particular clients or special clinical situations and cannot be considered inclusive of all practices or exclusive of other practices reasonably directed at obtaining the same results. In addition, the practice of genetic counseling is subject to regulation by federal, state and local governments. In a subject jurisdiction, any such regulations will take precedence over this statement. NSGC expressly disclaims any warranties or guarantees, express or implied, and shall not be liable for damages of any kind, in connection with the information set forth in this Scope of Practice statement or for reliance on its contents.

Genetic counseling is a dynamic profession, which undergoes rapid change with the discovery of new genetic information and the development of new genetic tests and treatment options. Thus, NSGC will periodically review and, where appropriate, revise this statement as necessary for consistency with current practice information."

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