

A Guide to Genetic Counseling

Second Edition

Edited by

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Division of Molecular Medicine and Genetics
University of Michigan

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*To our patients and their families, from whom we receive the
inspiration to pursue our work
and
to our students, who provide the
opportunity for us to look at our work in new ways.*

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Foreword

Genetic counseling is often thought of as a young profession. But that perception may be misguided. Genetic counseling as a clinical service was defined more than 60 years ago, the first genetic counseling class graduated nearly 40 years ago, the National Society of Genetic Counselors is 30 years old, and genetic counseling training programs span the globe. And now this book—the first comprehensive genetic counseling text—is in its second edition. We are not the new kids on the block anymore.

The appearance of youth is perhaps due to a widespread lack of awareness of the profession. Why would most people need to know about genetic services when such a narrow segment of the population initially needed our services?

Over the last four decades, the portion of the patient population that needs genetic counseling services has steadily expanded. When advances in genetic knowledge, theory, and technology inevitably found their way into the medical care and lives of families, a wider swath of the general public discovered anew the need for genetic counseling by properly trained professionals. In the early years, genetic counselors met mostly with parents of children with genetic diseases or adults who had relatively uncommon disorders like Neurofibromatosis, Marfan syndrome and Ehlers–Danlos syndrome. Then came pregnant women, at first, only those over 35, and eventually pregnancies with abnormal serum or sonographic findings and carriers of recessive diseases as carrier testing expanded to a wider range of conditions. This was followed by the 10% of breast/ovarian/colon cancer patients who have a hereditary basis to their disease. Now we are counseling patients and families who are at risk for cardiovascular, neurological, and other common diseases, as well as anyone with a computer who wants to utilize online genetic testing. We seem like a new profession to each segment of the population who suddenly need genetic services.

Genetic counseling is not primarily about diagnosing genetic diseases, although genetic counselors often play a key role in that process. Rather, genetic counseling is more about the steps leading up to deciding about a genetic test or what happens once a diagnosis is made or a test result is positive. This is where the human meets the genome, the stage on which the human drama—or comedy, as Balzac or Saroyan would broadly describe it—plays itself out as families learn to cope with and adapt to the medical, social, and psychological impact of hereditary diseases.

How do you tell a couple who have been trying to get pregnant for 10 years that their unborn baby—their one and only pregnancy—has a lethal condition? How do you break the news to immigrant parents, who barely speak English and are so proud of their first child born in a new country, that their son will be cognitively and physically challenged, without losing focus on the child’s humanity and dignity and the family’s cultural values? How do you discuss limiting physical activity with a 15-year-old star player of her school basketball team after a “fainting spell” leads to a diagnosis of long QT syndrome? How do you tell a newly married 30-year-old woman that she faces a high risk of developing ovarian cancer, the same disease her mother and sister died of by the time they were 40, while maintaining hope and guiding her to take control of her risk before her risk controls her? How do you explain to a 70-year-old man that his Parkinson-like symptoms are related to his grandson’s fragile X syndrome? What ethical principles guide you through these experiences? What counseling and educational skills do you need to help patients understand and act on their risks and diagnoses? How do you set up your clinic to make sure that your services are properly delivered and patients are appropriately evaluated and followed? How do you keep yourself educated in the rapidly expanding universe of genetic information? How do you maintain your sanity, health, commitment, and passion? How do you train the next generation of genetic counselors so they can use what we already know and adapt genetic counseling to discoveries about what we do not yet know?

Turn the page and find out.

*Swedish Medical Center
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ROBERT G. RESTA, M.A., M.S., C.G.C.

Preface

The first edition of this book was a landmark achievement—the first book written by genetic counselors on the principles and practice of genetic counseling. Since its publication in 1998, much has changed—the Human Genome Project has been completed, over 1700 genetic tests are available, and the profession has seen substantial growth, nationally and internationally. The goals of the second edition remain the same—to provide a comprehensive overview of genetic counseling, focusing on the components, theoretical framework, and unique approach to patient care that are the basis of this profession. The book covers the genetic counseling process, from case initiation to completion, and uses a broad lens that makes this information generally applicable to all areas of clinical genetic counseling.

Genetic counselors have created roles for themselves in an expanding array of medical specialties and assumed important roles in settings that extend beyond the clinic, including research, industry, commercial laboratories, Internet companies, public policy, education, and public health. In recognition of genetic counselors' role expansion, new chapters in this edition include “Genetic Counseling Research: Understanding the Basics,” “Evolving Roles, Expanding Opportunities,” and “Genetic Counselors as Educators.” In addition, there is a new chapter on “Risk Communication and Decision-Making.”

As in the first edition, the chapters are organized in a manner to facilitate academic instruction and skill attainment. Chapter 1 provides an overview of the history and field of genetic counseling. Chapters 2–11 describe the components of genetic counseling, including content on how to obtain family and medical history information, conduct a genetics medical evaluation, case preparation and management, genetic testing, medical documentation, patient interviewing, psychosocial considerations, multicultural counseling, patient education, risk communication and facilitation of

decision-making. Chapter 12 focuses on some of the ethical and legal issues that arise in cases and in our professional interactions with patients and other healthcare professionals. Chapters 13–17 address professional issues, which include providing and receiving clinical supervision, writing research proposals and conducting research, giving presentations, continuing education, professional development, and expansion of genetic counselors' positions in clinical and non-clinical settings. The final chapter helps the reader see how it all fits together, using specific genetic counseling case examples.

All chapters in the second edition have been updated. Case vignettes are included in several chapters, and there are several tables that can be used as checklists for case preparation and management, presentations, clinical supervision, writing research proposals, and conducting research. Several chapters are lengthy, packed with content and details, and will be best approached by reading in more than one sitting. You will find some topics addressed in more than one chapter, covered from different perspectives and for varied applications. Some topics are not covered in great depth because it would take an entire book to appropriately cover the topic. Given how rapidly web addresses change, we have kept website citations to a minimum, mainly those that cannot be accessed from a name search. Since the focus of this book is the practice of genetic counseling and not genetic conditions, we recommend that you access GeneReviews, Online Mendelian Inheritance in Man, and the literature for specific information about cited conditions.

The field of genetic counseling began in the United States and has spread internationally, with genetic counseling programs in many countries. We wish to acknowledge that our book is written from an American perspective; it is beyond the book's scope to address genetic counseling in other countries. The principles of genetic counseling are universal, even though the practice of genetic counseling varies nationally and internationally. Whether you are a student, teacher, supervisor, healthcare professional or just interested in learning about genetic counseling, this book will serve as a comprehensive introduction and reference.

We wish to acknowledge Diane L. Baker, M.S., C.G.C. for her leadership, vision, and dedication to the first edition of this book. We wish to express our appreciation to John Wiley & Sons—Blackwell Publishing for their commitment to publishing this and other books about genetic counseling, particularly Thomas Moore (Senior Editor, Medicine), Kristen Parrish (Production Editor), and Sanchari Sil (Senior Project Manager, Thomson Digital).

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1

The Practice of Genetic Counseling

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

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*

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 *ευγενής*, 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