

SECOND EDITION

GENETIC COUNSELING PRACTICE

ADVANCED CONCEPTS AND SKILLS

EDITED BY

BONNIE S. LEROY | PATRICA McCARTHY VEACH
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WILEY Blackwell

Genetic Counseling Practice

Genetic Counseling Practice

Advanced Concepts and Skills

Second Edition

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Preface

This edition, just like the first edition, is intended for genetic counseling students, practitioners, and educators. Although the focus is primarily on some of the more complicated issues that arise in the provision of genetic counseling services, the chosen topics will resonate well with anyone working in the helping professions. The genetic counselor authors share their extensive expertise from many areas of genetic counseling practice and research. Some are also cross trained in disciplines such as psychology and public health. This mix of expertise greatly enriches the ways in which we are able to understand our patients and families and further develop the skills that improve practice. Autonomy, patient decision-making, risk communication, grief, patient anger, cultural competency, resistance, and countertransference are a few of the counseling-centered topics covered in this text. These topics are universal to the care of all patients and families. Chapters on families, children and adolescents, and cultural diversity provide in-depth consideration of counseling patients of diverse backgrounds. We have also chosen to delve into the development of the genetic counseling professional with chapters on reflective practice and career development, and we look to the future through chapters describing how our skills will be used in new environments. Once again, the topics included in this text translate well to anyone working in health care settings providing genetic services to patients and families in any country around the globe. As this text is intended primarily for teaching, the authors have included examples that are concrete and drawn from real-life practice. Each chapter contains learning activities for use in the classroom. Many of the activities are suitable for practitioners to use individually, and/or in a peer supervision setting, for their professional growth.

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MaryAnn W. Campion, EdD, MS, LCGC received a BS in Psychology from Furman University and an MS in Genetic Counseling from the University of South Carolina. She has a doctorate in Educational Leadership and Policy from Boston University, where her dissertation focused on mid-career faculty vitality. She was the founding director of the Master's Program in Genetic Counseling and Assistant Dean in the Division of Graduate Medical Sciences at Boston University School of Medicine until 2016. Since that time, she has served as co-director of the Master's Program in Human Genetics and Genetic Counseling at Stanford University and Consultant on Genetic Counseling Professional Practice at Stanford Children's Health. Her previous clinical career centered on prenatal genetic counseling, working in that capacity for three years with the Greenwood Genetic Center and 12 years at Boston Medical Center, and she is now working in the Stanford Down Syndrome Clinic. Her research has concentrated on faculty development in academic medicine and the translation of genomics into public health. Her interest and expertise in genetic counselor education has been further enhanced by serving as a consultant on strategic planning and organizational analysis for genetic counseling training programs and through her roles as chair of the AGCPD New Program Development Sub-Committee and the ACGC Standards Committee.

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Acknowledgments

We were once again very fortunate to have been able to work with a group of experienced and talented authors who contributed their expertise in this second edition. We greatly appreciate their time and efforts which resulted in an edition that highlights critical topics supported by genetic counseling research. Please note that some chapters have been updated from the first edition by different authors. Most drew from the work of the authors in the first edition. We hope readers will enjoy learning from these chapters as much as we have.

Learn from yesterday, live for today, hope for tomorrow. The important thing is not to stop questioning.

Albert Einstein

1

Introduction

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I've learned that people will forget what you said, people will forget what you did, but people will never forget how you made them feel.

—Maya Angelou

This famous quote, frequently attributed to Maya Angelou, seems particularly meaningful as the genetic counseling profession grows in size and scope. The types of patients to whom genetic counselors are now able to offer services is increasing but the service delivery models are also changing. These changes bring exciting new opportunities as well as challenges. In the first edition of this book, published in 2010, we said, “The explosion of knowledge and multiple opportunities for patients to learn about their genetic risks have far out-paced advances in understanding complex psychosocial aspects of genetic counseling practice” (LeRoy et al. 2010, p. 1). Well, it is now clear, that was just the beginning. In 2010, direct to consumer (DTC) testing was in the initial stages of becoming available to the general population, and it would be three years before Angelina Jolie’s prophylactic mastectomy introduced countless people to the world of cancer genetics. Events such as these have contributed to making *genetics* a common term. Moreover, the growth of DTC testing has made searching for relatives using one’s own DNA, a popular pastime. *Genetics* now is not only a part of medicine identifying those at risk for disease and helping to direct the care of affected patients, it has a recreational side (e.g. using DNA to find relatives, learning about one’s ability to perform as an elite athlete, finding a mate). And, almost everyone has heard the word **genetics**.

Scientific advances tend to focus our attention on the rapid changes in information and technology. Genetic counselors do love the science of genetics, and for good reason – it is endlessly fascinating. Just when we think we understand something, new knowledge emerges that improves the way we understand the genetic contribution to disease and how we understand the very basis of disease itself. It’s easy (and even expected) to become fixed on the science; but it’s also important to remember that most patients are struggling to survive with a condition or help loved ones who are affected. Attending to this struggle is genetic counseling.

This edition, just as the first, reflects our belief that, although up-to-date, relevant genetic information is critical to the success of genetic counseling, the practice goes so much further. We said in the first edition, “genetic counselors are active and compassionate participants in patients’ efforts to understand their genetic risks, endure their emotional struggles, and make life-altering decisions” (LeRoy et al. 2010, p. 1) This is still true today as genetic counselors find many new venues in which to apply their skills and provide services.

As this edition goes to press, the National Society of Genetic Counselors is celebrating its 40th anniversary, and the first class of genetic counseling students entered Sarah Lawrence College 50 years ago. Literature specific to the practice of genetic counseling literature has grown. The *Journal of Genetic Counseling* serves as a major venue for disseminating genetic counseling research that helps to inform many areas of practice. Still, there are relatively few teaching texts that provide focused and detailed instruction in psychosocial methods of genetic counseling practice. This book addresses some of the more challenging areas of practice and provides strategies for addressing patient needs as well as promoting genetic counselor professional growth.

Our authors are an exceptional group. As experts in the profession, they draw from their diverse experiences in clinical, academic, and leadership settings to provide readers with relevant examples that bring key concepts and skills to life. Whenever possible, the authors cite relevant genetic counseling literature that provides an empirical foundation for their chapters. Each chapter also includes activities that promote teaching and learning of the concepts and skills. We thank the authors for their valuable contributions and their thoughtful work.

Development of the 2nd Edition

We based the choice of topics for the second edition on our belief that, for the most part, those areas covered in the first edition exemplify advanced skills that remain very relevant and would be enhanced with the inclusion of recent genetic counseling literature. We also included new topic areas that reflect the growth of the profession in new directions and have new literature to support this evolution. In this edition, we are purposeful in using the word *patient* rather than *client*. We believe individuals coming to genetic counselors for services, typically do so in a clinical setting and therefore see themselves as patients. Genetic counselors working in laboratories are also integral to directly or indirectly helping patients understand disease and make decisions that help them use genetics to their benefit. Genetic counseling is a health care profession. It is implied in our definition:

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:

- 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.

(Resta et al. 2006, p. 77)

Human behavior and the essence of genetic counseling practice have not changed, and so the Reciprocal-Engagement Model (REM) of genetic counseling practice (McCarthy Veach et al. 2007) continues to play a foundational role in this new edition.

Reciprocal-Engagement Model

The REM model is an amalgamation of data generated during a consensus meeting of North American genetic counseling program directors. Their task was to define a model of genetic counseling practice that was being taught at the time of the consensus meeting. A model of practice “refers to why and how the service is delivered to patients, as described by tenets, goals, strategies, and behaviors” (McCarthy Veach et al. 2007, p. 714). The participants worked to reach consensus regarding the tenets (fundamental beliefs shared by genetic counselors), goals (the aims of practice), strategies (methods for achieving goals), and behaviors (specific actions/interventions) that characterize genetic counseling practice. They identified five tenets.

- **Tenet 1: Genetic information is key** – Providing information about “perceived or known genetic contributions to disease” (p. 721) and engaging in discussion with patients about this information is a particularly unique aspect of genetic counseling.
- **Tenet 2: Relationship is integral to genetic counseling** – The quality of the relationship developed between the genetic counselor and patient is as important as genetic information. Genetic counseling “... is a relationally-based helping activity whose outcomes are only as good as the connection established between the counselor and patient” (p. 721).
- **Tenet 3: Patient autonomy must be supported** – Patients should be as self-directed as possible regarding genetic counseling decisions. The counselor is an active participant, working with the patient’s individual characteristics and family and cultural context to facilitate informed decisions. However, an essential aspect of this tenet is that “the patient knows best” (p. 721).
- **Tenet 4: Patients are resilient** – Most patients have the strength to deal with painful situations. Genetic counselors, therefore, encourage patients to draw upon their inner resources (coping strategies) and support systems/resources to make decisions and arrive at an acceptance of their situation.
- **Tenet 5: Patient emotions make a difference** – Patients experience a multitude of emotions that are relevant to genetic counseling. “Patient emotions interact with all aspects of genetic counseling processes and outcomes, for instance, affecting their desire for information, their comprehension of information, the impact of information on their decisions, their willingness and ability to connect with the counselor, their desire for autonomy, and their perceived resilience” (p. 722).

These five tenets are illustrated in a triangle that represents their mutual influence on each other (see Figure 1.1). The genetic counselor–patient relationship is in the center of the triangle because it comprises the conduit for the processes and outcomes of genetic counseling.

The chapters in the second edition of this book continue to correspond well to the REM. The relationship remains key as is evident in all chapters that speak to patient encounters. Authors provide readers with a deeper consideration of patient and counselor characteristics, genetic counseling relationship dynamics, and ways to enhance information provision. Also congruent with the REM, material included in this book addresses professional development and the necessity for genetic counselors to engage in life-long reflective practice. Reflection is essential for facilitating professional development and also for evaluating the impact of one’s service provision.

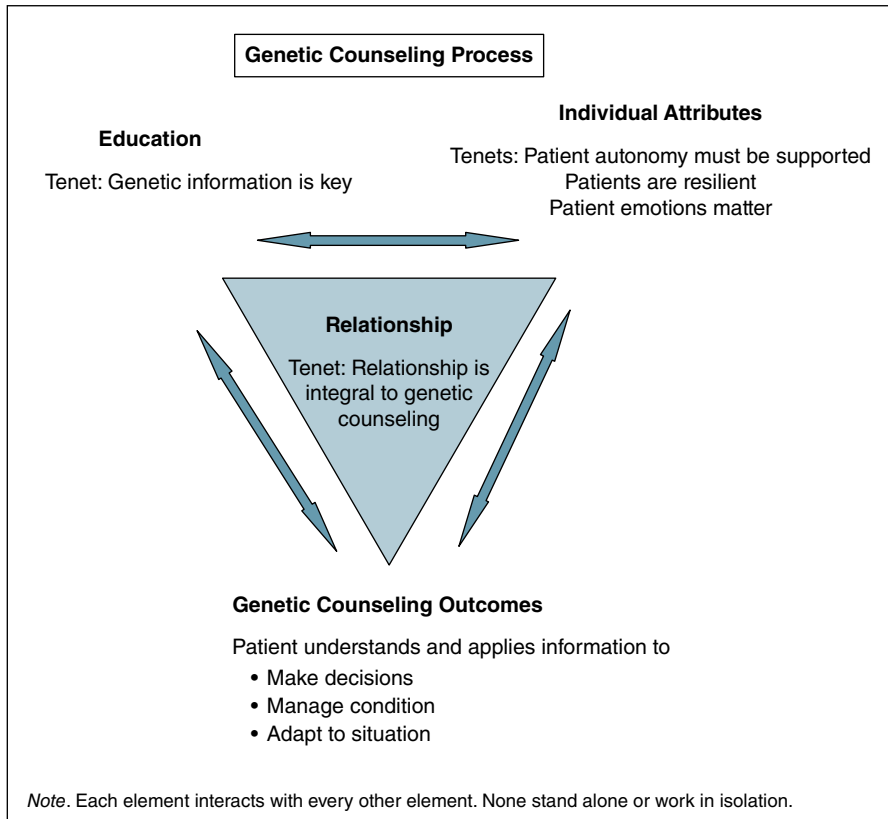


Figure 1.1 A reciprocal-engagement model of genetic counseling. Reprinted with permission from the Journal of Genetic Counseling.

Chapter Descriptions

The chapters in this addition address *universal* and enduring concepts that are relevant across different genetic counseling specialties and patient conditions. The chapters focus on some of the more challenging aspects of genetic counseling and stretch the counselor’s skills as they work to provide relevant information in ways that benefits patients and families.

Resta (Chapter 2: Complicated Shadows: The Limitations of Autonomy in Genetic Counseling Practice)

Resta again questions the place of autonomy in genetic counseling and what it means to really help patients. Resta points out that as a core value, autonomy is limited and sometimes may have a negative impact on genetic counseling. But as the REM describes, attending to other tenets as well, such as the importance of the relationship, are critical for the practice of genetic counseling (McCarthy Veach et al. 2007).

Shugar and Shuman (Chapter 3: Actively Engaging with Patients in Decision-Making)

Shugar and Shuman describe the historical transition in medicine to one where patients now play more of a collaborative role in the decisions that impact care. In addition, the authors illustrate some of the major barriers to decision-making and provide strategies for overcoming these barriers in the genetic counseling setting. They point out that facilitating decision making is a primary skill needed in many genetic counseling sessions and only through an understanding of the barriers that patients face, can the genetic counselor help patients make life changing choices.

Delaney Dixon, Campion, and Singletary (Chapter 4: Risk Communication: A Complex Process)

These authors take on one of the most challenging skills in genetic counseling practice. They help readers navigate through risk assessment, barriers to understanding risk, and strategies for communicating risk in a way that helps patients and families use genetic information to their benefit. A key tenet in the REM is that “Genetic Information is Key.” However, patients can only use this complex information if they are able to understand it and put it into perspective for their own situation. This chapter helps readers to develop the skills needed to effectively communicate risk.

Bellcross and Ali (Chapter 5: Grieving: An Inevitable Journey), and Schema (Chapter 6: Facing Patient Anger) and Redlinger Grosse (Chapter 7: Resistance and Adherence: Understanding the Patient’s Perspective, and Chapter 8: Countertransference: Making the Unconscious Conscious)

These chapters tackle some of the most challenging psychosocial aspects of genetic counseling, involving times in the session when the counselor can feel as overwhelmed as the patient. Each of these topics is highly emotional and typically tap into the counselor’s own personal reactions. With reflective practice, the counselor can effectively address these challenges in the moment to more effectively help patients. These authors provide readers with a better understanding of not only what a patient might be experiencing but also, what the counselor is experiencing and how the counselor can more effectively provide support and help patients make sense of the disorder they and their families are facing.

McAllister, Metcalfe and MacLeod (Chapter 9: Supporting Communication about Genetic Conditions in a Family) and Austin and Schoch (Chapter 10: Developmentally Based Approaches for Counseling Children and Adolescents)

These chapters remind readers that when providing genetic counseling services, counselors are working within the framework of a family. The decisions patients make, the developmental constraints on patient decisions, and the impact of the information counselors provide are all influenced by the patient and their family in some way. These authors provide excellent strategies for understanding and helping patients within the context of their family.

Warren (Chapter 11: Cultural Competency and Genetic Counseling: Key Concepts) and (Chapter 12: Cultural Competency: Application to Genetic Counseling)

Warren provides key concepts and related skills drawn from her years of experience researching and publishing on cultural competency and its relevance to genetic counseling practice. In her first chapter, Warren describes the critical relevance of cultural competency to genetic counseling practice and helps readers explore personal biases that often are barriers to effective genetic counseling. In the second chapter, she challenges readers to work toward developing the skills needed to provide culturally competent

genetic counseling. She states, “Every genetic counselor has a professional responsibility to engage in a cultural competency journey to more effectively meet the needs of a culturally diverse patient” (Chapter 12, p. 263).” As our profession grows and the ability to provide genetic services to a greater diversity of patients increases, so does our responsibility to ensure that all patients and use genetics to their health benefit.

Durst and Mills (Chapter 13: Health Literacy Skills)

This is one of the new chapters in the second edition of this book. We added this chapter because of its salience to genetic counseling processes and outcomes, and because of a growing literature that supports the importance for health care providers to better understand how people learn. It is critical for genetic counselors to have the skills needed to translate complicated information so patients and families can use that information to their benefit. These skills are emphasized in the Practice Based Competencies that are expected of every genetic counselor. Two of the genetic counseling practice-based competencies that form the core skills needed for practice in genetic counseling involve the skill of educating:

“Domain III: Education

14. Effectively educate clients about a wide range of genetics and genomics information based on their needs, their characteristics and the circumstances of the encounter.

15. Write concise and understandable clinical and scientific information for audiences of varying educational backgrounds.”

(Accreditation Council for Genetic Counseling 2015)

Trepanier and Allain (Chapter 14: Applying Genetic Counseling Skills in Evolving Environments), Baty (Chapter 16: Genetic Counseling Career Development), and Reiser (Chapter 15: Growing as a Professional and the Role of Reflective Practice)

The authors of these chapters remind readers that the genetic counseling profession is ever evolving. Trepanier and Allain illustrate many ways to provide genetic counseling services so more patients and families can have access. The strategies they describe reflect both the profession’s dedication to providing services to as many patients as possible and genetic counselors’ creativity and flexibility. Reiser articulates the necessity of reflective practice as a means for personal growth when talking about genetic counselors’ professional journey, “not because there is an end to the journey but rather because a professional journey requires continual review and revision” (Chapter 15, p. 342). In the final chapter, Baty brings the history of our profession to life demonstrating how genetic counselors’ understanding of this history can illuminate the future. Baty challenges readers to become active in the genetic counseling profession, not only to promote their own growth but also to become drivers for future directions of the field.

Considerations

We encourage readers to critically reflect upon the concepts and strategies presented and determine how they fit their personal style and practice needs. Furthermore, some of the case examples, while clearly illustrating counseling skills, are necessarily brief and thus cannot capture the full context of a patient–counselor encounter. The examples and corresponding strategies are intended to serve as guides for

practice and not as formulas for service provision and professional development. Importantly, all cases and examples presented in the chapters have been anonymized.

As in the first edition, each of the chapters concludes with several learning activities to facilitate skill development. Most of the activities are intentionally interactive so students and/or practitioners can learn from one another, and most activities can be modified for groups or individual learning. Although, this text and the activities are directed toward more advanced genetic counseling students who have had some clinical exposure, the material and activities would also be useful for supervisors working with advanced students, genetic counselors in peer supervision groups, and genetic counseling practitioners who are interested in refreshing their skills.

In Closing

We want to express heartfelt thanks to our authors who contributed to this edition. We have learned a great deal from such an experienced group of professionals, and we are grateful to them for sharing their expertise! We hope you will enjoy reading these chapters as much as we have, and we wish everyone the personal and professional satisfaction that comes from working in such a great profession.

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2

Complicated Shadows: The Limitations of Autonomy in Genetic Counseling Practice

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OBJECTIVES

- Define autonomy and its role in genetic counseling practice
- Identify limitations of autonomy as a primary guiding ethical principle in genetic counseling practice
- Identify social, cultural, and economic factors that can influence the ability of patients to act autonomously
- Develop ethically sound counseling strategies for situations in which patient autonomy may be not desired by the patient or reliance on autonomy may not be in the patient's best medical or psychosocial interests

*There's a line you must toe
But it's darker than you know
In those complicated shadows.*

—Elvis Costello, “Complicated Shadows”

Sheldon Reed defined genetic counseling in 1947 (Resta 1997), coincidentally the same year that the Nuremberg Code on human experimentation was formalized by a US military court in *United States of America v. Karl Brandt, et al.* The Nuremberg Code has profoundly influenced medical ethics, particularly the development of individual autonomy as the primary guiding principle of medical ethics in general (Fischer 2006) and of genetic counseling specifically.

Although the Nuremberg Code does not contain the word “autonomy,” its first item is a clarion call for autonomy in medical experimentation:

“The voluntary consent of the human subject is absolutely essential. This means that the person involved should have legal capacity to give consent; should be so situated as to be able to exercise free power of choice, without the intervention of any element of force, fraud, deceit, duress,

over-reaching, or other ulterior form of constraint or coercion; and should have sufficient knowledge and comprehension of the elements of the subject matter involved as to enable him to make an understanding and enlightened decision.”

(<https://history.nih.gov/research/downloads/nuremberg.pdf>)

In the decades after the *Trials of War Criminals before the Nuremberg Military Tribunals* the US and much of Western Europe witnessed social changes such as increased reproductive freedom, the growth of patient advocacy, and the decline of medical paternalism, all of which further contributed to autonomy assuming a central ethical role in society and in medicine. Genetic counseling – and the genetic counseling profession – had their origins in these times. The first genetic counseling program was established at Sarah Lawrence College in 1969, and the first class of master’s degree level genetic counseling students graduated in 1971. This was contemporaneous with the growth of the modern women’s rights movement in the US as well as the 1973 *Roe v. Wade* decision, which legalized abortion in the US. Initially, most genetic counseling jobs were in prenatal diagnosis or pediatric clinics and focused on reproductive aspects of medical genetics. Thus, it is not surprising that the genetic counseling profession embraced a patient-centered (i.e. female) ethos that encouraged individual choice (Stern 2012).

Although one presumed effect of the increased availability of abortion after the *Roe v. Wade* ruling was a decrease in the number of pregnancies, actually the 1980s witnessed a dramatic increase in the number of pregnancies to women 35 and older, concomitant with the growth of the genetic counseling profession in the US and most of Western Europe (Resta 2005). This surge in pregnancies, primarily among educated higher income women, provided economic and employment justification for the fledgling genetic counseling profession and also provided an opportunity for autonomy to become the bioethical rallying cry of genetic counselors when amniocentesis became more widely available in the late 1970s and early 1980s (Stern 2012). Even in 2019, when genetic counseling has expanded to many specialties beyond prenatal diagnosis, autonomy remains a core component of genetic counseling ethos and practice. Autonomy was initially understood and articulated within the context of reproductive autonomy, but it is now more widely incorporated into all areas of genetic counseling that involve patient choice (e.g. patients who undergo whole exome sequencing and decide not to learn the risk of developing certain diseases, or BRCA mutation carriers who are considering risk-reducing mastectomy).

Definition and Focus

Autonomy has been defined in many ways with differing emphases and nuances. Based on clinical experience, discussions with colleagues, and extensive reading of the genetic counseling literature, it is my impression that genetic counselors usually view non-coercive non-directiveness as a means of supporting autonomy. Therefore, within the context of genetic counseling, I define autonomy as *the principle that individuals should make medical and reproductive decisions free from physical, social, political, economic, and psychological constraints*. The emphasis in this definition is on the individual, although in practice autonomy may extend beyond an individual. Autonomy also includes situations like a couple trying to make a joint decision about a child or a pregnancy or a family trying to make a decision about