

SECOND EDITION

# GENETIC

## COUNSELING PRACTICE

### ADVANCED CONCEPTS AND SKILLS

EDITED BY

BONNIE S. LEROY | PATRICA MCCARTHY VEACH  
NANCY P. CALLANAN



WILEY Blackwell

# Table of Contents

[Cover](#)

[Title Page](#)

[Copyright Page](#)

[Preface](#)

[About the Authors](#)

[Acknowledgments](#)

[1 Introduction](#)

[Development of the 2nd Edition](#)

[References](#)

[2 Complicated Shadows: The Limitations of Autonomy  
in Genetic Counseling Practice](#)

[Definition and Focus](#)

[Seven Limitations of Autonomy](#)

[Summary](#)

[Learning Activities](#)

[References](#)

[3 Actively Engaging with Patients in Decision-Making](#)

[A Brief History of the Medical Encounter: From  
Paternalism to Shared Decision-Making](#)

[Psychological Aspects of Decision-Making](#)

[Informed Decision-Making](#)

[Summary](#)

[Learning Activities](#)

[Acknowledgment](#)

[References](#)

## 4 Risk Assessment and Communication: A Complex Process

Introduction

Risk Assessment in Genetic Counseling

Risk Communication

Influences on Risk Perception

Summary

Learning Activities

References

## 5 A Genetic Counselor's Guide to Understanding Grief

Introduction

The Grief Process

Types of Grief

Conclusion

Learning Activities

Appendix 5.A Additional Examples of Anticipatory Guidance for Patients

Appendix 5.B Unhelpful and Helpful Things to Say to Grieving Patients

References

## 6 Patient Anger: Insights and Strategies

Introduction

Challenges Posed by Patient Anger

Practice Applications

Conclusion

Summary Points

Learning Activities

References

## 7 Resistance and Adherence: Understanding the Patient's Perspective

Definition and Concepts

Resistance

Adherence and Non-adherence

Summary

Learning Activities

## 8 Countertransference: Making the Unconscious Conscious

Definitions and Concepts

The Countertransference Process

Manifestations and Effects of Countertransference

Management of Countertransference

Summary

Learning Activities

References

## 9 Supporting Family Communication About Genetic Conditions

Introduction

Literature Review

Application of Select Family Systems Theory

Interventions Adapted to Genetic Counseling

Summary

Learning Activities

Acknowledgments

References

## 10 Developmentally Based Approaches for Counseling Children and Adolescents

Introduction

[Child and Adolescent Development](#)

[Theoretical Framework](#)

[Chronic Illness and Children](#)

[Strategies for Genetic Counseling with Children and Adolescents](#)

[Conclusion](#)

[Summary Points](#)

[Learning Activities](#)

[References](#)

## [11 Cultural Competency and Genetic Counseling: Key Concepts](#)

[Introduction](#)

[Definitions and Key Concepts](#)

[Health Disparities](#)

[Conceptualizing Cultural Competence](#)

[Developing Cultural Competency](#)

[Summary](#)

[Learning Activities](#)

[Resources for Cultural Knowledge, Cases, Teaching, and Learning Exercises](#)

[Further Reading](#)

[Websites](#)

[References](#)

## [12 Cultural Competency: Application to Genetic Counseling](#)

[Introduction](#)

[Cultural Competency and the Genetic Counseling Process](#)

[Explanatory Models of Health and Well-being](#)

[Health Literacy](#)

[Working with Interpreters and Translators](#)

[Summary](#)

[Learning Activities](#)

[Resources for Cultural Knowledge, Cases, Teaching, and Learning Exercises](#)

[References](#)

### [13 Education in Genetic Counseling: The Impacts of Genetic Literacy and Adult Learning](#)

[Introduction](#)

[General Literacy in the United States](#)

[Health Literacy](#)

[Genetic Literacy](#)

[Genetic Literacy Considerations for Genetic Counselors](#)

[Adult Education Theory in Practice](#)

[Summary Points](#)

[Learning Activities](#)

[References](#)

### [14 Adapting Genetic Counseling Practice to Different Models of Service Delivery](#)

[Introduction](#)

[Defining Service Delivery Models](#)

[Attributes of Alternative Service Delivery Models](#)

[Putting it all Together - Practice Implications](#)

[Conclusions](#)

[Learning Activities](#)

[References](#)

## 15 “Oh The Places You’ll Go!” The Genetic Counselor Professional Development Journey

Introduction

Literature Review

Reflective Practice

Optimizing Self-Reflection

Leadership

Summary Points

Acknowledgments

Appendix 15.A Case Discussion Template

References

## 16 Development of the Genetic Counseling Profession: A Professionalization Process

What Makes a Field of Practice a Profession?

How Does Genetic Counseling Measure Up to the Characteristics of a Profession?

Milestones in the Development of the Genetic Counseling Profession

Genetic Counseling and the General Model of Professionalization

Looking to the Future: Milestones that Need Further Development to Advance the Professionalization of Genetic Counseling

How is Professionalization of Genetic Counseling Relevant to Genetic Counseling Practitioners?

Summary

Learning Activities

References

Index

End User License Agreement

# List of Tables

## Chapter 3

[Table 3.1 Integrative model of shared decision making.](#)

[Table 3.2 Genetic counseling approaches to support shared decision making.](#)

## Chapter 4

[Table 4.1 Bayes theorem: applied to clinic.](#)

[Table 4.2 Bayes theorem applied to case 1.](#)

[Table 4.3 Positive Predictive Value \(PPV\) in a testing situation.](#)

[Table 4.4 Clinical example for PPV.](#)

[Table 4.5 Bayes theorem for residual risk after negative NIPT for 30 y/o woma...](#)

[Table 4.6 Using Bayes theorem to incorporate reproductive history.](#)

[Table 4.7 Using Bayes theorem to assess carrier risk based on family and repr...](#)

[Table 4.8 Common heuristics.](#)

[Table 4.9 Biases that may alter risk perception.](#)

## Chapter 5

[Table 5.1 Comparison of grief and major depression.](#)

[Table 5.2 Concepts and phrases helpful for pediatric genetic sessions.](#)

## Chapter 10



[Table 10.1 Children's common coping strategies and illustrative examples.](#)

## Chapter 13

[Table 13.1 ACGC practice-based competences for genetic counselors \(ACGC 2015\)...](#)

[Table 13.2 Words scored on the short 8-item REALM-R health literacy tool \(Bas...](#)

[Table 13.3 Words included on the short 8-item REAL-G genetic literacy assessm...](#)

[Table 13.4 Summary of common readability tools.](#)

[Table 13.5 Assessment tools for patient materials.](#)

[Table 13.6 Simplified words for use in written materials \(CDC n.d.; Universit...](#)

[Table 13.7 Knowles concept of learners in applied practice.](#)

[Table 13.8 Kessler's two models of genetic counseling.](#)

[Table 13.9 2018 PSS teaching/education audiences \(National Society of Genetic...](#)

## Chapter 14

[Table 14.1 Description of the most common genetic counseling service delivery...](#)

[Table 14.2 Benefits and limits of common service delivery models.](#)

## Chapter 15

[Table 15.1a Targeted rubric-based evaluation tool \(incorporating specific che...](#)

[Table 15.1b Practice-based competencies and milestones \(reprinted from Guy et...](#)

[Table 15.2 Examples of RIME structure applied to genetic counseling practice ...](#)

## Chapter 16

[Table 16.1 Milestones in the professionalization of the genetic counseling pr...](#)

[Table 16.2 Trends in techniques for increasing diversity in genetic counselin...](#)

# List of Illustrations

## Chapter 1

[Figure 1.1 A reciprocal-engagement model of genetic counseling. Reprinted wi...](#)

## Chapter 3

[Figure 3.1 Three talk model of shared decision making.](#)

## Chapter 4

[Figure 4.1 Impact of severity on risk. \\*\\*Larger % of liability under the cur...](#)

## Chapter 5

[Figure 5.1 Elizabeth Kübler-Ross' original stages of grief model.](#)

[Figure 5.2 Modified Kübler-Ross seven-stage model.](#)

[Figure 5.3 The grieving wheel.](#)

## Chapter 9

[Figure 9.1 Simplified genogram shows both the family pedigree but also notes...](#)

## Chapter 13

[Figure 13.1 Oral literacy demand framework.](#)

[Figure 13.2 Strategies for clear communication and encouraging patient engag...](#)

## Chapter 15

[Figure 15.1 Advanced training for certified genetic counselors: An overview ...](#)

## Chapter 16

[Figure 16.1 Genetic counseling professionalization process.](#)

# Genetic Counseling Practice

## Advanced Concepts and Skills

Second Edition

*Edited by*

***Bonnie S. LeRoy, MS, CGC***

*Professor*

*Department of Genetics, Cell Biology and Development*

*University of Minnesota*

*Minneapolis, MN, USA*

***Patricia McCarthy Veach, PhD, LP***

*Professor Emerita*

*Department of Educational Psychology*

*University of Minnesota*

*Minneapolis, MN, USA*

***Nancy P. Callanan, MS, CGC***

*Director Emeritus*

*MS Genetic Counseling Program*

*University of North Carolina at Greensboro*

*Greensboro, NC, USA*

**WILEY** Blackwell

This edition first published 2021  
© 2021 John Wiley & Sons Ltd.

*Edition History*

Wiley-Blackwell (1e, 2010)

All rights reserved. No part of this publication may be reproduced, stored in a retrieval system, or transmitted, in any form or by any means, electronic, mechanical, photocopying, recording or otherwise, except as permitted by law. Advice on how to obtain permission to reuse material from this title is available at <http://www.wiley.com/go/permissions>.

The rights of Bonnie S. LeRoy, Patricia McCarthy Veach, and Nancy P. Callanan to be identified as the author(s) of the editorial material in this work has been asserted in accordance with law.

*Registered Office(s)*

John Wiley & Sons, Inc., 111 River Street, Hoboken, NJ 07030, USA  
John Wiley & Sons Ltd, The Atrium, Southern Gate, Chichester, West Sussex, PO19 8SQ, UK

*Editorial Office*

9600 Garsington Road, Oxford, OX4 2DQ, UK

For details of our global editorial offices, customer services, and more information about Wiley products visit us at [www.wiley.com](http://www.wiley.com).

Wiley also publishes its books in a variety of electronic formats and by print-on-demand. Some content that appears in standard print versions of this book may not be available in other formats.

*Limit of Liability/Disclaimer of Warranty*

The contents of this work are intended to further general scientific research, understanding, and discussion only and are not intended and should not be relied upon as recommending or promoting scientific method, diagnosis, or treatment by physicians for any particular patient. In view of ongoing research, equipment modifications, changes in governmental regulations, and the constant flow of information relating to the use of medicines, equipment, and devices, the reader is urged to review and evaluate the information provided in the package insert or instructions for each medicine, equipment, or device for, among other things, any changes in the instructions or indication of usage and for added warnings and precautions. While the publisher and authors have used their best efforts in preparing this work, they make no representations or warranties with respect to the accuracy or completeness of the contents of this work and specifically disclaim all warranties, including without limitation any implied warranties of merchantability or fitness for a particular purpose. No warranty may be created or extended by sales representatives, written sales materials or promotional statements for this work. The fact that an organization, website, or product is referred to in this work as a citation and/or potential source of further information does not mean that the publisher and authors endorse the information or services the organization, website, or product may provide or recommendations it may make. This work is sold with

the understanding that the publisher is not engaged in rendering professional services. The advice and strategies contained herein may not be suitable for your situation. You should consult with a specialist where appropriate. Further, readers should be aware that websites listed in this work may have changed or disappeared between when this work was written and when it is read. Neither the publisher nor authors shall be liable for any loss of profit or any other commercial damages, including but not limited to special, incidental, consequential, or other damages.

*Library of Congress Cataloging-in-Publication Data*

Names: LeRoy, Bonnie S., editor. | Veach, Patricia McCarthy, editor. | Callanan, Nancy P., editor.

Title: Genetic counseling practice : advanced concepts and skills / edited by Bonnie S. LeRoy, Patricia McCarthy Veach, Nancy P. Callanan.

Description: Second edition. | Hoboken, NJ : Wiley-Blackwell, 2021. | Includes bibliographical references and index.

Identifiers: LCCN 2020028243 (print) | LCCN 2020028244 (ebook) | ISBN 9781119529859 (paperback) | ISBN 9781119529811 (adobe pdf) | ISBN 9781119529866 (epub)

Subjects: MESH: Genetic Counseling | Professional-Patient Relations

Classification: LCC RB155.7 (print) | LCC RB155.7 (ebook) | NLM QZ 52 | DDC 362.196/042-dc23

LC record available at <https://lcn.loc.gov/2020028243>

LC ebook record available at <https://lcn.loc.gov/2020028244>

Cover Design: Wiley

Cover Images: DNA structure © Mit apevski/Shutterstock, Business team © Jolkesky/Getty Images

## 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.

*Bonnie S. LeRoy*

*Patricia McCarthy Veach*

*Nancy P. Callanan*

## About the Authors

**Nadia Ali, PhD** is Co-Assistant Director of Emory University's Genetic Counseling Training Program, where she trains future genetic counselors in the counseling skills necessary for compassionate, effective patient care. She is also Director of Psychological Resources for Emory's Genetic Clinical Trials Center, in which she conducts clinical research into neurocognitive and psychological manifestations of Lysosomal Storage Diseases and other metabolic disorders, as well as coordinating psychometry for clinical drug trials. Dr. Ali earned her doctorate in Clinical Psychology from the University of South Florida and completed an internship in Neuropsychology and a post-doctoral fellowship in Health Psychology.

**Dawn C. Allain, MS, LGC** is an Associate Professor in the Department of Internal Medicine; Director of Genetic Counseling Graduate Program; and Associate Director of the Division of Human Genetics at The Ohio State University. She received her MS in Genetic Counseling from Northwestern University in 1993, and she is a diplomate of the American Board of Genetic Counseling. She provides genetic counseling services to patients in the Adult Genetics and Genomics Program, as well as the Nisonger Center's Interdisciplinary Developmental Clinic. Her research interests focus on genetic counseling service delivery models, genetic counselor credentialing, genetic services billing and reimbursement, and genetic counselor and health professional education.

**Stephanie Austin, MS, MA, CGC** is a practicing Genetic Counselor/Senior Research Program Leader at Duke University in Durham, NC where she has worked since 2007. In her clinical time, she works in a metabolic clinic

setting. Her research focus is lysosomal storage diseases, glycogen storage disorders, and new treatments for rare disease. She is cross trained in genetic counseling and counseling psychology.

**Bonnie Jeanne Baty, MS, CGC, LGC** is Professor Emerita in the Department of Pediatrics and Program Director Emerita of the Graduate Program in Genetic Counseling at the University of Utah. She established genetic counseling services at the University of Utah and has authored more than 40 journal articles and book chapters. In her emerita life, she continues to teach genetic counseling students, conduct and publish research in genetic counseling education and professional development, and serve on multiple national committees and as editor for a genetic counseling book series for Oxford University Press. She is a recipient of the Natalie Weissberger Paul lifetime achievement award and remains active in the National Society of Genetic Counselors, founding and co-chairing the Late Career Special Interest Group.

**Cecelia Bellcross, PhD, MS, CGC** is an Associate Professor with the Department of Human Genetics at Emory University School of Medicine. She obtained both her Master of Science degree in Medical Genetics and her PhD in Population Health Sciences through the University of Wisconsin-Madison. Following 16 years in clinical practice, she completed a post-doctoral fellowship at the CDC's Office of Public Health Genomics. She developed and launched the Emory Genetic Counseling Training Program in 2012.

**Nancy P. Callanan, MS, CGC** is Director Emeritus of the MS Genetic Counseling Program at the University of North Carolina at Greensboro. She is a past president of the National Society of Genetic Counselors and the American Board of Genetic Counseling and was the recipient of the

Natalie Weissberger Paul National Achievement Award in 2008. She had over 20 years of clinical experience prior to establishing the genetic counseling program at UNCG and serving as the first program director. She is co-author of the 2018 textbook, *Facilitating the Genetic Counseling Process*.

**MaryAnn W. Champion, 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.

**Shannan DeLany Dixon, MS, CGC** is the Director of the Master's in Genetic Counseling (MGC) Training Program at the University of Maryland School of Medicine where she

also serves as an Assistant Professor in the Department of Pediatrics. She received her BS in Biology with a concentration in Genetics from Penn State University and an MS in Genetic Counseling from Arcadia University. Prior to serving as the director of the MGC Program she worked in a variety of settings providing pediatric, cancer, and prenatal genetic counseling services to multiple diverse patient populations. Her most recent area of research interest involves medical transition and the development of tools to assist with this complex process. She is grateful to have multiple opportunities to work with the greater genetic counseling community in several leadership positions including, most recently secretary treasurer of the National Society of Genetic Counselors (NSGC) and Vice President of the Association of Genetic Counseling Program Directors (AGCPD).

**Andrea L. Durst, MS, DrPH, LCGC** is the Associate Director of the Genetic Counseling Program and the Co-Director of the MPH Program in Public Health Genetics at the University of Pittsburgh where she teaches courses on both genetic counseling and public health genetics. She has eight previous years of experience in cancer genetic counseling. Dr. Durst is currently working with a number of organizations to advance public health genetics projects. She serves as Chair of the Steering Committee for the New York Mid-Atlantic Regional Genetics Network (NYMAC), as Secretary/Treasurer of the Pennsylvania Association of Genetic Counselors (PAGC), and is a previous Co-Chair of the National Society of Genetic Counselors (NSGC) Public Health Special Interest Group. She has previously provided her public health genetics management expertise to projects with the Midwest (Region 4) Genetics Network and The Genetic Alliance.

**Bonnie S. LeRoy, MS, LGC** is a Professor in the Department of Genetics, Cell Biology, and Development,

former Director of the Graduate Program of Study in Genetic Counseling and Associate Member of the Center for Bioethics at the University of Minnesota. Bonnie is also a member of the Academy of Distinguished Teaching Professors at the University of Minnesota. She is a past president of the National Society of Genetic Counselors and the American Board of Genetic Counseling and she was the recipient of the Natalie Weissberger Paul National Achievement Award in 2003. She completed two terms as the Editor-in-Chief of the *Journal of Genetic Counseling* from 2008 to 2018.

**Rhona MacLeod, BN, MSc, PhD, RGN** is a Consultant Genetic Counselor at Manchester Centre for Genomic Medicine (MCGM). Rhona completed a Bachelor of Nursing degree at Glasgow University, MSc (Psychiatry) and PhD at Manchester University. She is Lead for the STP MSc Genomic Counseling programme at the University of Manchester. Rhona's clinical interests relate to Neuro-genetics and new models of developing support for families following genetic and genomic testing. She is Co-leader of the European Huntington's Disease Network Working Group "Genetic Counseling and Testing." Since 2015, Rhona has been working with Clinical Psychologist Mariangels Ferrer-Duch to develop Narrative group sessions for patients known to the genetic family register service at MCGM.

**Marion McAllister, PhD** is a registered genetic counselor (UK Genetic Counsellor Registration Board and European Board of Medical Genetics), with a PhD in Social Science from Cambridge University (1999), and over 10 year's clinical experience. Marion is currently Programme Director for the MSc in Genetic and Genomic Counseling at Cardiff University. She has a special interest in patient empowerment, and a distinguished track record in genetic counseling research. Marion's research aims to improve

how clinical genetics services respond to the needs of families with genetic conditions, including supporting family communication about genetic conditions. She developed and validated a Patient Reported Outcome Measure (Genetic Counseling Outcome Scale GCOS-24) for clinical genetics services, which captures patient benefits conceptualized as patient empowerment. Since its publication in 2011, the GCOS-24 has been translated into Dutch, Danish, Portuguese, Spanish, and Japanese and is being used to evaluate clinical genetics services in practice, and new interventions in research in the UK and internationally.

**Patricia McCarthy Veach, PhD, LP** is a licensed psychologist and an Emerita Professor at the University of Minnesota. She is a member of the Academy of Distinguished Teachers at the University and recipient of a Minnesota Psychological Association Outstanding Graduate Faculty in Psychology Award. Her teaching and research involve supervision, training, and professional development of genetic counselors and other human services professionals. She has authored three books and over 100 professional articles. She served as Assistant Editor of the *Journal of Genetic Counseling* from 2008 to 2018.

**Alison Metcalfe, PhD** is Professor of Health Care Research and Pro Vice Chancellor for Health and Wellbeing. She is an academic researcher who is a registered nurse, with a PhD in immunology and biophysics and a postgraduate certificate in Family Therapy. She has worked with families for almost 30 years. Alison's research focuses on co-designing interventions with families to facilitate communication about genetic conditions affecting them, which improves their resilience and ability to adapt to living with the disease and its associated risks for future generations.

**Rachel Mills, MS, LCGC** is an Assistant Professor with the Genetic Counseling Program at the University of North Carolina Greensboro. There, she serves as an instructor and the Research and Capstone Coordinator. She joined the UNCG faculty in August of 2019 after working in clinical research at Duke University for 11 years. Her research interests and specialization include precision medicine, pharmacogenetics, ethical, legal and social issues in genetics, and genetics education. She is an active volunteer with NSGC, serving on the Annual Conference Program Planning committee, Education Committee and various SIGs. Ms. Mills received her MS in Genetic Counseling from UNCG and is currently pursuing a doctorate in Adult Education from North Carolina State University

**Krista Redlinger-Grosse, PhD, LP, ScM, LGC** received her master's in genetic counseling from the JHU/NIH Genetic Counseling Training Program and PhD in Counseling Psychology from the University of Minnesota. She is on faculty with the University of Minnesota Graduate Program in Genetic Counseling as an Assistant Professor and Associate Director of Clinical Training. She also works as a psychologist in private practice providing assessment and psychotherapy services. Her interests center around the integration of the fields of genetic counseling and psychology through clinical work, education/training, and research.

**Catherine A. Reiser, MS, CGC** is the Program Director of the Master of Genetic Counselor Studies at the University of Wisconsin-School of Medicine and Public Health. She is also a practicing clinical genetic counselor with over 30 years of experience in pediatric and prenatal settings. Casey has served as past president of the Association of Genetic Counseling Program Directors and on committees and task forces for the NSGC and ACGC. She has received both the Audrey Heimler Award (Development of a Training



Program for Clinical Supervisors Using the Standardized Student Method) and the Jane Engleberg Memorial Fellowship (Development of Educational Resources for Genetic Counseling Students, Genetic Counselors and Students in other Health Professions: Digital Recordings of Genetic Counseling Sessions Conducted by Master Genetic Counselors).

**Robert G. Resta, MS, CGC** is a genetic counselor with 36 years of clinical experience in prenatal testing and hereditary cancers. He has a long-standing interest in the ethical, historical, and social implications of genetic counseling and genetic testing, and is a former editor-in-chief of the *Journal of Genetic Counseling*. He is a frequent contributor to *The DNA Exchange* blog, and a co-author of the textbook *Advanced Genetic Counseling - Theory and Practice*.

**Lynn Schema, MS, CGC** is a practicing pediatric genetic counselor at the University of Minnesota Medical Center - Fairview. She primarily specializes in metabolic and lysosomal diseases, as well as cystic fibrosis. Lynn is a graduate of the University of Minnesota Genetic Counseling Program, and currently she serves as a clinical supervisors and lecturer for the Genetic Counseling graduate program.

**Kelly Schoch, MS, CGC** is a practicing Genetic Counselor at Duke University in Durham, NC where she has worked since 2008. She has provided research and clinic coordination for the Duke 22q11.2 Deletion Syndrome Clinic. For the past 10 years she has worked primarily in a research setting focused on patients with undiagnosed diseases undergoing genomic sequencing.

**Andrea Shugar, MS, CGC** is a Genetic Counsellor in the Department of Genetic Counselling at The Hospital for Sick Children where she sees children and families for a wide

variety of conditions. She has been instrumental in the creation of a number of multidisciplinary clinics, most notably for individuals with 22q deletion syndrome and their families. Andrea is past president of the Canadian Association of Genetic Counsellors. She is a Lecturer in the Molecular Genetics department at the University of Toronto, supervising clinical learning and research along with teaching advanced genetic counselling skills in MSc Program in Genetic Counselling. Andrea has published on clinical care, genetic counselling skills and professional regulation.

**Cheryl Shuman, MS, CGC** is Director of the Department of Genetic Counselling at the Hospital for Sick Children. She is also the Program Director of the MSc Program in Genetic Counselling where she teaches and provides clinical and research supervision and is a Professor in the Molecular Genetics department at the University of Toronto. Cheryl's research interests include clinical and (epi)genetic correlations (i.e. overgrowth syndromes with cancer predisposition) and the provision of genetic and genomic counselling (e.g. clinical and personal utility of genome-based testing technologies and inherent ethical issues).

**Claire N. Singletary, MS, CGC** is the Director of the Genetic Counseling Program at the University of Texas MD Anderson UTHealth Graduate School of Biomedical Sciences and an Associate Professor in the Departments of Pediatrics and of Obstetrics, Gynecology and Reproductive Sciences at McGovern Medical School. She received her BS in Psychology from Furman University and her MS degree in Genetic Counseling from Northwestern University. Prior to directing the program at UTHealth, Claire was the Assistant Director at the University of South Carolina Genetic Counseling Program, where she also provided prenatal genetic counseling services. She remains

passionate about practicing the art of prenatal genetic counseling today. In addition, she provides didactic instruction in several psychosocial courses, professional development courses, and the prenatal genetic counseling course. Her main research interests are in risk perception, new prenatal technologies, and psychosocial counseling. She is active in the National Society of Genetic Counselors, the Texas Society of Genetic Counselors, and the AGCPD, where she served most recently as Vice President.

**Angela Trepanier, MS, CGC** is a Professor in the Center for Molecular Medicine and Genetics and Director of the Genetic Counseling Graduate Program at Wayne State University. She received her MS in Genetic Counseling from the University of Minnesota and is a diplomate of the American Board of Genetic Counseling. In addition to her work with the genetic counseling program, Ms. Trepanier works with the Michigan Department of Health and Human Services to improve appropriate access to genetic counseling and testing for hereditary cancer syndromes through provider and patient education efforts. Her research interests include investigating utilization of genetic counseling service delivery models, mechanisms for improving access to genetic counseling, psychosocial aspects of genetic counseling, and genetic counseling outcomes.

**Nancy Steinberg Warren, MS, CGC**, is owner of Genetic Counseling Toolkit, LLC. She provides education and training for practicing genetic counselors and students in cultural competency, as well as consulting for institutions of higher learning on the design and curriculum of established and emerging genetic counseling programs. She was awarded the 2009 NSGC Jane Engelberg Memorial Fellowship to support her work in understanding the challenges and opportunities of minority recruitment and cultural diversity in the field of genetic counseling. As an

outcome of the Fellowship, she developed an online resource portal and continuing education course for genetic counselors, A Genetic Counseling Cultural Competence Toolkit

<http://www.geneticcounselingtoolkit.com/>. This resource is widely used by genetic counselors and graduate programs. In 2013, Ms. Warren was the first recipient of the NSGC Cultural Competence Award. For most of her professional career, Ms. Warren's focus has been on genetic counseling program development and evaluation, and teaching and mentoring students. She has been a Program Director and faculty member at the University of Cincinnati and Cincinnati Children's Hospital, Long Island University-CW Post, and Bay Path University Genetic Counseling Graduate Programs.

## 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

*Bonnie S. LeRoy<sup>1</sup>, Patricia McCarthy Veach<sup>1</sup>, and Nancy P. Callanans<sup>2</sup>*

*<sup>1</sup> Department of Genetics, Cell Biology and Development, University of Minnesota, Minneapolis, MN, USA*

*<sup>2</sup> University of North Carolina at Greensboro, Greensboro, NC, USA*

*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