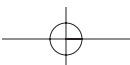
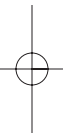
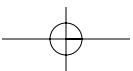
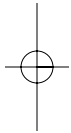
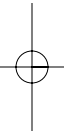


Psyche and Helix





Psyche and Helix

Psychological Aspects of Genetic Counseling

Essays by Seymour Kessler, PhD

Edited by Robert G. Resta, MS

Division of Perinatal Medicine
Swedish Medical Center
Seattle, Washington

 **WILEY-LISS**

A JOHN WILEY & SONS, INC., PUBLICATION

NEW YORK • CHICHESTER • WEINHEIM • BRISBANE • SINGAPORE • TORONTO

This book is printed on acid-free paper. ∞

Copyright © 2000 by Wiley-Liss, Inc. All rights reserved.

Published simultaneously in Canada.

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, scanning or otherwise, except as permitted under Sections 107 or 108 of the 1976 United States Copyright Act, without either the prior written permission of the Publisher, or authorization through payment of the appropriate per-copy fee to the Copyright Clearance Center, 222 Rosewood Drive, Danvers, MA 01923, (978) 750-8400, fax (978) 750-4744. Requests to the Publisher for permission should be addressed to the Permissions Department, John Wiley & Sons, Inc., 605 Third Avenue, New York, NY 10158-0012, (212) 850-6011, fax (212) 850-6008, E-Mail: PERMREQ@WILEY.COM.

For ordering and customer service, call 1-800-CALL-WILEY.

Library of Congress Cataloging-in-Publication Data:

Psyche and helix : psychological aspects of genetic counseling / edited by Robert G.

Resta ; essays by Seymour Kessler.

p. cm.

Includes index.

ISBN 0-471-35055-9 (alk. paper)

1. Genetic counseling—Psychological aspects. I. Kessler, Seymour. II. Resta, Robert G.

RB155.7 .P78 2000

616'.042—dc21

00-020717

Printed in the United States of America.

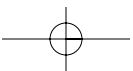
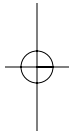
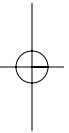
10 9 8 7 6 5 4 3 2 1

To Susan, Emily, and Lizzie.

—RR

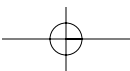
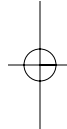
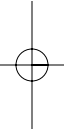
To my grandchildren, who speak for the future.

—SK



Contents

Preface <i>Robert G. Resta</i>	ix
1. Analysis of a Transcript	1
2. Quantitative Analysis of a Transcript of a Genetic Counseling Session	19
3. Management of Guilt and Shame	35
4. Suffering and Countertransference	63
5. Preselection: A Family Coping Strategy in Huntington Disease	68
6. Advanced Counseling Techniques	73
7. More on Counseling Skills	85
8. Empathy and Decency	99
9. A Critical Review of the Literature Dealing with Education and Reproduction	109
10. Teaching and Counseling	135
11. Thoughts on Directiveness	142
12. Nondirectiveness Revisited	150
13. Notes and Reflections	165
Index	173
	<i>vii</i>



Preface

Genetic counseling has been defined in many ways since Sheldon Reed first coined the phrase more than 50 years ago (Resta, 1997). Depending upon the training and philosophical bent of the practitioner, genetic counseling has been viewed as an exercise in risk calculation, psychosocial assessment, education about medical and genetic information, a communication process, or a euphemism for eugenics. In some ways, all of these definitions are correct. After all, genetic counseling is defined by the practice of professionals who call themselves genetic counselors. Genetic counseling *is* what genetic counselors *do*.

So what should a genetic counselor *do* during a counseling session? Counselees almost always answer that question. Sometimes we can simply ask clients what they want from us (Michie et al., 1997). Often, though, we must rely on clinical skills and experience to elicit from patients what they want to gain from their time with us. Whatever agenda the counselor brings to the table, what matters most is what is important to counselees (Michie et al., 1998). If clients are not interested in discussing Bayesian risk calculation, population improvement, their innermost feelings, DNA mutations, or whatever a counselor *believes* is important to patients, then the session is for naught (Berkenstadt et al., 1999; Shankar et al., 1999). Everybody loses. Clients come away frustrated, their needs unmet (Freyer et al., 1999; McGowan, 1999). Counselors are left puzzled about why they cannot get through to counselees and become annoyed at the latter's inability to understand technical information despite the counselor's valiant efforts to explain it.

One way to help assure a rewarding counseling experience is to dispel the notion that clients who come for genetic counseling are primarily seeking education. This is not to say that clinical and educational issues are not important to them (Smith, 1998). Indeed, receiving a diagnosis and having the diagnosis and testing options explained are very important aspects of client satisfaction (Berkenstadt et al., 1999;

x PREFACE

McCarthy Veach et al., 1999; Michie et al., 1997). But if education is the primary goal of genetic counseling, then a well-written book, Internet site, or a CD-ROM could better serve patients.

I argue that genetic counseling is more than an exercise in specialized education. At its core, genetic counseling is about helping people try to understand and cope with the effects of genetic disease on their lives and the lives of their families (Street and Soldan, 1998). Genetic counseling touches on the emotions and behaviors that define us as human—suffering, sacrifice, reproduction, self-image, family relationships, guilt, joy, anger, denial, love, and the struggle to comprehend the unpredictable world in which we live (Taswell and Sholtes, 1999). Indeed, genetic counseling is a *de facto* psychological process (Reif and Baitsch, 1985; Shiloh, 1996). A client's understanding of, and reaction to, any information we provide is shaped by a complex filter composed of the client's unique psychological makeup, emotional status, age, sex, culture, and developmental and personal experiences (Finucane, 1998; Lippman, 1999). As much as we might want to believe otherwise, counselees do not plan a perfectly logical course of reproductive or medical action based on a mathematical weighing of risks and advantages. People are not, in the words of songwriter Tom Waits, "bone machines."

Because genetic counseling can raise so many psychological issues, should genetic counselors be psychotherapists? No, at least they should not be psychotherapists *sensu strictu*. Most genetic counselors are neither trained as psychotherapists, nor do most genetic counseling patients suffer from psychopathology. Nevertheless, good counseling requires the counselor to have certain basic counseling skills:

- The ability to understand the psychological needs of others.
- The ability to understand the psychological meaning of clients' behaviors.
- The ability to communicate that understanding in ways that leave clients emotionally enriched, psychologically stronger, and more competent to deal with their own lives.

We also need to recognize that the very act of genetic counseling may result in a psychotherapeutic experience for the client. Learning about the cause of a child's mental retardation may help alleviate the guilt of a mother who had blamed her child's problems on a medication she took while she was pregnant, or some kind words from a counselor offered at the appropriate time during a session may help a patient come to terms with his or her disorder (Targum, 1981; Eunpu, 1997; Matloff, 1997; Resta, 1998; Tuttle, 1998; Biesecker and Marteau, 1999; Peters et al., 1999). But many patients who seek genetic counseling are not in need of the long-term specialized care of a psychotherapist, or should be referred to a psychotherapist when appropriate.

While genetic counselors need not be full-time psychotherapists, they do need to demonstrate basic counseling skills to achieve the goals of genetic counseling. A genetic counselor must be able to calculate risks, provide clear and accurate information about complex medical issues, and possess good administrative skills. But even the most complex risk calculation is a useless statistic if the counselor does not possess

the ability to help the client put that risk in a meaningful and appropriate psychological context. What does it matter if a 35-year-old woman has “only” a 5% risk for developing ovarian cancer if the counselor does not understand that the woman has refrained from having children because she would not want her young children to watch her die from this disease at a young age, just like she watched her own mother die 20 years ago? A counselor might not understand a woman’s ambivalence about undergoing amniocentesis unless the counselor knows that as a child the woman mercilessly teased the neighbor boy who was mentally retarded, and feels that she should be “punished” by making her first-born child have Down syndrome.

There are many excellent books and articles about clinical genetics and the calculation of recurrence risks. There are very few publications that provide practical and clinically meaningful ways to enhance the counseling skills of genetic counselors. Dr. Kessler’s essays form the largest systematic approach to understanding and applying basic counseling skills to the practice of genetic counseling. Published over two decades in *American Journal of Medical Genetics* and *Journal of Genetic Counseling*, the essays retain their crispness and relevance to the daily practice of genetic counseling.

Many people contributed to the development of this book and the essays and we are most grateful to them, especially Dr. John Opitz, Dr. Charles Epstein, Pat Ward, Hilda Kessler, A.G. Jacopini, Dr. Alan Leveton, Colette Bean and Luna Han at John Wiley and Sons, Inc., Carol Prince, and Kluwer Academic/Human Sciences Press.

REFERENCES

- Berkenstadt M, Shiloh S, Barkai G, Katznelson M B-M, Goldman B (1999) Perceived personal control (PPC): a new concept in measuring outcome of genetic counseling. *Am J Med Genet* 82:53–59.
- Biesecker BB, Marteau TM (1999) The future of genetic counseling: an international perspective. *Nat Genet* 22:133–137.
- Eunpu D (1997) Systematically-based psychotherapeutic techniques in genetic counseling. *J Genet Counsel* 6:1–20.
- Finucane B (1998) *Working with Women Who Have Mental Retardation*. Elwyn, PA: Elwyn, Inc.
- Freyer G, Dazord A, Schlumberger M, Conte-Devoix B, Ligneau B, Trillet-Lenoir V, Lenoir GM (1999) Psychosocial impact of genetic testing in familial medullary-thyroid carcinoma: a multicentric pilot-evaluation. *Ann Oncol* 10:87–95.
- Lippman A (1999) Embodied knowledge and making sense of prenatal diagnosis. *J Genet Counsel* 8:225–274.
- Matloff ET (1997) Generations lost: a cancer genetics case report. *J Genet Counsel* 6:169–186.
- McCarthy Veach P, Truesdell SE, LeRoy BS, Bartels DM (1999) Client perceptions of the impact of genetic counseling. *J Genet Counsel* 8:191–216.
- McGowan R (1999) Beyond the disorder: one parent’s reflection on genetic counselling. *J Med Ethics* 25:195–199.
- Michie S, Marteau TM, Bobrow M (1997) Genetic counselling: the psychological impact of meeting patients’ expectations. *J Med Genet* 34:237–241.

xii PREFACE

- Michie S, Weinman J, Marteau T (1998) Genetic counselors' judgments of patient concerns: concordance and consequences. *J Genet Counsel* 7:219–232.
- Peters JA, Djurdjinovic L, Baker D (1999) The genetic self: The Human Genome Project, genetic counseling and family therapy. *Fam Syst Health* 17:5–25.
- Reif M, Baitsch H (1985) Psychological issues in genetic counselling. *Hum Genet* 70:193–199.
- Resta R (1997) Sheldon Reed and fifty years of genetic counseling. *J Genet Counsel* 6:375–378.
- Resta R (1998) Carolyn's feet. *Am J Med Genet* 72:1–2.
- Shankar A, Chapman P, Goodship J (1999) Genetic counseling: do we recognise and meet the consultands' agenda? *J Med Genet* 36:580–582.
- Shiloh S (1996) Genetic counseling: a developing area of interest for psychologists. *Prof Psychol Res Prac* 27:475–486.
- Smith ACM (1998) Patient education. In: Baker D, Schuette JL, Uhlmann WR (eds). *A Guide to Genetic Counseling*. New York: Wiley-Liss, pp 99–126.
- Street E, Soldan J (1998) A conceptual framework for the psychosocial issues faced by families with genetic conditions. *Fam Syst Health* 16:217–232.
- Targum S (1981) Psychotherapeutic considerations in genetic counseling. *Am J Med Genet* 8:281–289.
- Taswell HF, Sholtes SK (1999) Predictive genetic testing: a story of one family. *Fam Syst Health* 17:111–121.
- Tuttle LC (1998) Experiential family therapy: an innovative approach to the resolution of family conflict in genetic counseling. *J Genet Counsel* 7:167–186