

## Book Review

COUNSELING IN MEDICAL GENETICS, THIRD EDITION, Sheldon Reed, Alan R. Liss, Inc., New York, 1980, 246 pp., \$26.00.

A book about genetic counseling by the man who coined the term in 1947 is difficult to criticize. Founders have the right to define their disciplines, much as a chef can stick by a recipe. We may prefer other methods but we cannot dispute authenticity.

The approach Dr. Reed takes toward genetic counseling is authentic and genuine. Chapter 3 is a delightful combination of wit and wisdom that would forearm anyone undertaking this surprisingly complex task of counseling families. He defines genetic counseling as "a kind of social work . . ." whose major function is "to provide people with an understanding of the genetic problems in their families." The primary requirement for diagnosis ("the soft underbelly of medical practice") and willingness of the counselor to listen and communicate at the patient's level are emphasized. The counselor must be nondirective (he "does not drive the family car for them, he rides along with them and shares the view"), and he must aggressively explore the burden that a risk figure has for the family. The "bright light" in counseling is that most families have a pessimistic view of their situation, and even the clumsiest counselor has a good chance of relieving anxiety.

Despite this excellent beginning, two problems beset the remaining chapters. One is a matter of emphasis—the choices between general principles and detailed material are not consistent. Another involves perspective—certain diseases, practices, and professional roles that inevitably concern modern genetic counselors are not considered. I suspect that both problems relate to the explosive increase in scientific information and services that has occurred in medical genetics. A thorough treatment of genetic counseling may be beyond the reach of a single author volume. Chapter 5, for example, discusses chromosomal disease without providing guiding principles of dysmorphology or details of cytogenetic methodology. Rather than suggesting multiple anomalies or mental retardation as indications for karyo-

typing, cytogenetic screening of all children is stated as a goal that awaits more efficient technology. Criteria for karyotypic evaluation in reproductive failure, sexual ambiguity, or X-linked mental retardation are never mentioned, and the details about sex chromosome anomalies seem misplaced in the absence of a section on Down syndrome.

Chapter 8, which outlines Mendelian patterns of inheritance, also omits important concepts. Some introduction to statistics and probability would be helpful for later chapters on multifactorial inheritance. Especially useful would be the relation of segregation patterns to a binomial distribution and the generation of Gaussian curves for traits determined by multiple genes. No mention of gene action is made, thus missing a chance to use blood genetics (Chapter 10) as an example of molecular defects that can be classified as dominant or recessive according to the precision of molecular analysis. Certain clinically useful generalizations, such as the greater severity of recessive forms of a disease, or the fact that recessive diseases tend to be metabolic and dominant disease structural, are never mentioned. Even the practical dilemmas of counseling—the investigation of variably expressive dominant disease, the detection of carriers for X-linked disease—are not illustrated with examples of case material.

Other gaps include the approach to treatable metabolic disorders in Chapter 7, where no mention of the usefulness of a urinary metabolic screen is made. Certain counseling roles, such as that of the genetics associate, are also omitted. These masters-degree-trained individuals have made a tremendous impact on many genetics clinics and screening programs by their focus on patient understanding and follow-up, issues that may be neglected by medical personnel. Indeed, the whole issue of how state services and counseling clinics should be organized is skirted, except to laud the early involvement of lay and state public health groups in Minnesota. I also take strong issue to statements in Chapters 18 and 22 that excuse the counselor from medical decisions such as heart surgery in Down syndrome. Even if the counselor lacks medical training,

he or she may give consolation and support to the parents during such crises by reaffirming genetic and prognostic information. Such crises emphasize the separate roles of supportive and informative counseling.

A strong point of the book is the coverage of twins, multifactorial inheritance, and normal traits in Chapters 14–17. Methods for the assessment of the hereditary and environmental components of many complex diseases are presented in very clear fashion. Empirical data are cited for diseases as diverse as cancer and pyloric stenosis, although more frequent use of tables might have made the data more accessible. This approach is extended to mental retardation, the psychoses, and certain environmental agents in Chapters 18–20.

In summary, Dr. Reed gives us an excellent philosophy for counseling and many useful but somewhat inaccessible empiric risk figures for common diseases. His concept of a genetic counselor seems somewhat limited and approaches to dysmorphology, connective tissue disease, neuromuscular disease, and metabolic screening are not mentioned at all. The book is an excellent introduction to the subject of genetic counseling but suffers as a reference. A redeeming feature, however, is the judgement and experience of the author, which shines through most pages and makes up for many

omissions. A paragraph worth quoting in these troubled times comes from Chapter 6 on amniocentesis: "A frequent question is when does life begin? Life does *not* begin with the fertilization of the egg. It began millions of years ago and has continued ever since then. It never starts anew. Both the egg and the sperm are living material, as is obvious to anyone who has seen them through the microscope. How long each person's life lasts is entirely conditional upon the genotype of the person and the environment it experiences. Death can occur at any time and is the usual fate of all unfertilized eggs and sperms. All life is an extension of pre-existing and continuing life. The egg has the potentiality of *becoming* a human being after elaborate developmental changes have occurred. But as a simple human cell it has none of the attributes of a human *being* and does not deserve to be considered a human being. The question becomes, 'at what point does this blob of cells *become* a human being?' The answer is a purely arbitrary determination, which results from one's philosophy."

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