Book Reviews

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Principles and Practice of Medical Genetics. 3d ed. Edited by David L. Rimoin, J. Michael Connor, and Reed E. Pyeritz. Senior Advisory Editor, Alan E. H. Emery. Secaucus, NJ: Churchill Livingston, 1996. Pp. 2937. $310.00.

This two-volume text, now in its third edition, is an impressive compendium of current knowledge in the rapidly evolving field of medical genetics. This edition was ably compiled by two of the same editors, David L. Rimoin and J. Michael Connor, who presided over the first and second editions. Reed E. Pyeritz joins them as a third editor, and Alan E. H. Emery remains as senior advisory editor. The editors are also exceptional contributors to many of the book's entries.

The dramatically increased of this edition scope mirrors our expanding molecular knowledge of the pathogenesis of disease states. Weighing in at 6.6 kg (vs. 4.5 kg for the 1990 edition), the text now has 235 contributors and 142 chapters. The organization of this edition remains similar to earlier versions, with 23 chapters devoted to basic principles and the remainder to clinical applications. A particularly personal and appealing new chapter, written by Victor A. McKusick, appropriately begins this work with the history of medical genetics. Many of Dr. McKusick's personal photographs, collected over a lifetime, illustrate and underscore the distinguished heritage of medical genetics. Pertinent new chapters in the section "Approaches to Clinical Problems" include those on infertility, mental retardation, and transplantation genetics. Another addition, the exhaustive and comprehensive chapter by Douglas Wallace, Michael Brown, and Marie Lott, "Mitochondrial Genetics," reviews an emerging field important to not only pediatric geneticists and neurologists but also those concerned with problems of later onset neurological deterioration and common manifestations of aging. Almost without exception, the entries are detailed and current and attempt to focus the reader on reliable concepts and principles without excessive attention to minutia. There is some unavoidable repetition in the various authors' choice of examples and photographs. The overall lack of repetitive written text is a tribute to the editors' zeal and skill.

There is something here to interest graduate and undergraduate students of medical genetics as well as nearly anyone practicing medicine today. In addition to the chapters devoted to general and clinical principles and categorical approaches to conditions such as short stature, there are multiple entries on specific systems (gastrointestinal, endocrine, and so on) and overviews of disease groups such as the muscular dystrophies, epilepsies, and chondrodysplasias. Adult common disease states including asthma, hypertension, diabetes mellitus, hemochromatosis, and the cardiomyopathies, are ably covered. Rapid progress in the understanding of molecular genetics and its consequent implications for patient and population screening, as well as disease detection and treatment, will likely demand more space in the future.

Genetic forms of cancer and familial predisposition to cancer are handled under each specific system (e.g., retinoblastoma under ophthalmologic disorders and breast cancer under endocrinologic disorders). Our increasing knowledge of the genetic basis of cancer will also require more comprehensive treatment and a separate overview chapter in subsequent editions.

This edition of *Principles and Practice of Medical Genetics* is a welcome addition to the truly outstanding reference texts in medical genetics today. It joins the ranks of *Mendelian Inheritance in Man, The Metabolic and Molecular Bases of Inherited Disease,* and *Recognizable Patterns of Human Malformation* and is a historical landmark with respect to the state of genetic knowledge in 1996. Certainly, readers will need to consult recent literature and on-line Web sites when researching a specific disorder, but the basic principles and most of the specifics should stand until the anticipated next edition after the year 2000. According to the publisher, a CD-ROM version of these two volumes will be available in late summer of 1997. This will be a boon in portability and rapid information access for those at home or in outside clinics. *Principles and Practice of Medical Genetics* belongs in every medical and college library and will be an essential addition to the personal libraries of practicing medical ge-

neticists. Accolades to the contributors and editors of this outstanding work!

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Optimizing Genetics Services in a Social, Ethical, and Policy Context: Suggestions from Consumers and Providers in the New England Regional Genetics Group (NERGG). By Dorothy C. Wertz and Robin Gregg. *The Genetic Resource,* Special Issue, Volume 10, Number 2, 1996. $10.00.

The active New England Regional Genetics Group has produced a detailed document describing ideal interactions between consumers and providers in the provision of genetic services. The project included contributions from five focus groups: three groups of consumers (from three different states), one of M.D./Ph.D. geneticists, and one of M.S. genetic counselors. The project team does not intend for this to be a "standard-of-care" document. The goal is that this document "could serve as a resource and reference for persons wishing to improve various components of genetic services" (p. 2).

The document is divided into three parts. Part A, "Approaches to Consumer-Provider Interactions," emphasizes the information obtained from the consumer groups and the consumer members of the project team. This section first discusses general considerations for these relationships, such as respect for persons, preserving family integrity, and the responsibilities of both the professional and the consumer. The needs and experiences of consumers are then listed as a message to providers. Part A concludes with an acknowledgment of the need for the general public to have some basic genetics education and with information that a consumer should know about a genetics referral.

Part B, "Social, Ethical, and Policy Contexts of Genetic Services," is the largest of the three parts, with 16 sections containing detailed information about a variety of issues. A team approach was an important consideration for consumers and is described first. Screening versus testing (including testing children), informed consent, and confidentiality are entries one would expect. There are also entries on parenthood for persons with disabilities, what consumers need to know when participating in research projects, and the danger of stigmatization involved with behavioral genetics. The uses and ethical limitations of cost-benefit analysis come next, followed by a discussion of the roles of the genetics center and the public health department.

Part C, "Specific Points for Direct Interactions," again uses the contributions of the focus groups for a more clinical approach to the preceding information. This part addresses optimal genetic counseling, the need to recontact patients, presymptomatic and susceptibility testing, various settings such as family planning clinics or pediatric clinics where genetic issues arise, adoption, prenatal diagnosis, and abortion.

This work represents thoughtful, detailed information that can be of use in a variety of ways. Examples of focused applications would be use of the section on duty to recontact as the basis of a review of policy in one's own genetics group or the use of the information contained in some of the many tables to make slides for a presentation enhancing genetics knowledge. Broader applications will include the use of the information included for education and policy decisions that will enhance health care in general and genetics health care in particular.

Copies of this document may be obtained from Joseph Robinson, MPH, Coordinator, New England Regional Genetics Group, P.O. Box 670, Mt. Desert, ME 04660 (telephone: [207] 288-2704; fax: [207] 288-2705; e-mail: 76363.3114@compuserv.com) at a cost of $10.

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