Book Reviews

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Principles and Practice of Medical Genetics. 2d ed. Edited by Alan E. H. Emery and David L. Rimoin. Edinburgh, London, Melbourne, and New York: Churchill-Livingstone, 1990. Pp. 2035. $255.00.

Two major questions come to my mind when faced with reviewing this massive two-volume set: (1) Do these volumes fill a particular need in the medical genetics library? and (2) Is the new edition sufficiently changed from the previous edition to be worth the expensive reinvestment? In answer to the first question, I believe that such a compendium of medical genetics is especially useful to fellows in training. These two volumes cannot take the place of the McKusick (1990) *Catalogue* for frequently updated information on a condition; but the McKusick *Catalogue* is not the place to find a scholarly and integrated discussion of the multiple aspects of a genetically inherited disease. On the other hand, although the two volumes under review have over 2,000 pages of text, this is not sufficient space for the authors to be as thorough as other monographs dealing with particular subtopics, e.g., dysmorphic syndromes, for which the Gorlin et al. (1990) monograph *Syndromes of the Head and Neck* provides far greater detail on almost every syndrome. For those of us who have been in the field of medical genetics for a period of time, our personal reprint collections are likely to be more up-to-date than this volume. Thus, in answer to question two, I would have to wonder whether the reinvestment is justified. Many chapters have no references past 1987, and others have relatively few references past that time. Given the many recent molecular advances that are dramatically altering the face of our subspecialty, timeliness is crucial, and these volumes do not do well in that regard. In fact, Victor McKusick's comment in his forward—i.e., that "the second edition of *Principles and Practice of Medical Genetics* is marked particularly by advances in this area of clinical molecular genetics"—is not supported by my analysis of the two volumes.

The monograph starts with more than 200 pages devoted to basic principles, much of which is excellent. In this section the chapters on "Teratogenic Agents" (a thorough review by James W. Hanson but without mention of modern molecular mechanisms), a "Clinical Approach to the Dysmorphic Child," and "Short Stature" are not to be expected in a human genetics textbook and are useful introductory material. I can't resist pointing out that the excellent chapter entitled "Gene Structure and Function in Eukaryotic Organisms" perpetuates the myth that "UAA is sometimes described as *amber* and UAG or UGA as *ochre* because of the color produced by bacterial strains which harbor mutant tRNAs capable of pairing with these termination codons." Actually, the *amber* mutants were named after R. H. Epstein's mother, and the *ochre* suppressors were named to keep up the sequence of colors (Hayes 1967, p. 422).

The section on "Basic Principles" is followed by about 100 pages on cytogenetics. The chapters are mostly thorough and useful. Lubs provides an excellent overview of X-linked mental retardation syndromes but could not include the recent, exciting molecular advances on the fragile X. The chapter by Chandley on "Infertility and Recurrent Abortion" is a thoughtful overview of a subject which sometimes generates controversy. However, it, too, contains few recent references.

Most of the rest of the two volumes is arranged by organ and functional systems. The section on "Neurological Disorders" starts with a very complete overview of genetics in prevention of neurotube defects and "uncomplicated" hydrocephalus by K. M. Lawrence. It is an exceptionally well-illustrated chapter. The section on "Mental Disorders" includes an excellent chapter on schizophrenia and the major

mood disorders, while the chapter on addictive disorders is less well written. I am at a loss to know why the case-oriented review on congenital myopathies is included in this section rather than in the following section on "Neuromuscular Disorders." "Ophthalmologic Disorders" and "Hereditary Deafness" are surveyed, and Drs. Cohen, Fraser, and Gorlin do a yeoman's job in managing to describe "Craniofacial Disorders" in less than 50 pages by providing massive tables of syndromes. Each entry in their table is a syndrome and not a single case, as was true in the chapter on "Congenital Myopathies." "Dermatological Disorders," "Skeletal Disorders," and "Connective Tissue Disorders" are given ample coverage, as are the other organ systems. "Immunological Disorders," "Endocrinological Disorders," and "Metabolic Disorders" receive their own subsections—but obviously the metabolic disorders are not nearly as completely covered as they are in *The Metabolic Basis of Inherited Disease* (Scriver et al. 1989). A single chapter on "Pharmacogenetics" and two covering "Neoplastic Disorders" complete this section. I was pleased to see in this last subsection a chapter on oncogenes, but it is very brief compared with the profusion of exciting new information in this field, much of it relevant to cancer genetics (treated in a separate chapter).

The book ends with a section on "Applied Genetics," which provides brief overviews of "Genetic Counseling" (a chapter that only skims the psychosocial issues), a quite thorough review of "Newborn Genetic Screening," and separately, "Prenatal Diagnosis and Therapy." "Genetic registers" and "Heterozygote Screening" are covered in addition to the "Treatment of Inherited Metabolic Diseases," which includes a quite up-to-date review of somatic gene therapy by Resnick. Brief chapters on "Paternity Testing" and on "Legal Considerations in the Delivery of Genetic Care" are followed by a final chapter on "Challenges for the Future." It is not surprising that it is hard to provide a capping chapter for a 2,000 page book, and many of the future considerations mentioned have already been surpassed by molecular genetic developments in the interval from the submission of the chapter to this reviewer's reading of the book. The book ends with a very thorough and obviously highly useful index (which is conveniently replicated in both volumes).

In conclusion, a detailed comparison of the first and second editions shows that the greater part consists of chapters on the same subjects by the same authors. When these rewritten chapters are contrasted between the two editions, they are seen to be mostly only slightly or moderately changed. The occasional new reference has been added, and topics have been slightly expanded, but the most recent advances in the molecular biology of the diseases are not included. Although the changes are for the better, they are not of sufficient magnitude to warrant purchasing the second edition if you have the first edition. Perhaps the changes are best compared with those to be expected in successive editions of standard clinical medicine textbooks which are purchased by the new arrivals (mostly medical students) but which established practitioners less frequently exchange for previous editions. They are certainly very different than the quite marked changes which usually occur between editions of *The Metabolic Basis of Inherited Disease* (Scriver et al. 1989).

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The Chromosomes in Human Cancer. 2d ed. By Avery A. Sandberg. New York, Amsterdam, and Oxford: Elsevier, 1991. Pp. xxviii + 1315. $265.00.

This long-awaited revision, after 10 years, of an important reference work is a boon to the working cytogeneticist and is a useful specialized resource for students, clinicians, and researchers. It is not a cozy volume to read in bed on a cold winter's night: of the 1,315 pages, 269 pages are devoted to bibliography, and there are innumerble tables, diagrams, illustrations, and other aids to the text. Its value as a referral source is enormous; as an introduction to critical principles and the role of cytogenetics within the broader context of tumor biology, it is less successful.

It is remarkable that such an immense text retains a highly personal quality; Dr. Sandberg is explicit that the contents reflect his own views as well as his wide experience and collaborations. His view of the evolution of the field must be respected, but, in places, his personal philosophy transcends a balanced perspective. Some controversial and important topics, such as the role of chromosome aberrations