Book Reviews

Am. J. Hum. Genet. 76:368, 2005

Inborn Errors of Development: The Molecular Basis of Clinical Disorders of Morphogenesis. Edited by Charles J. Epstein, Robert P. Erickson, and Anthony Wynshaw-Boris. New York: Oxford University Press, 2004. Pp. 1,082. $265.

Embryology and dysmorphology have evolved from descriptive sciences to molecular genetics endeavors. This book is first in making the leap from a classic catalog of malformations, syndromes, sequences, and associations to an organized approach to molecular analysis of patterns of morphogenesis. As we discover more about the intricate pathways of development, I expect any future editions of this text to expand to two or more volumes, similar to the expansion of *The Metabolic and Molecular Bases of Inherited Disease.*

Most clinicians in genetics have a deep curiosity about how our patients developed the medical problems that bring them to us. We identify syndromes and then attempt to explain the pathogenesis and etiology to the families. This task is formidable, and a resource such as this book can assist clinicians in organizing an approach to diagnosis by use of developmental pathways. In this book, the presentations are clear, the figures are extremely well done, and the references are complete.

There are a few topics that I would encourage the editors to include in a next edition. For example, there is no mention of Marfan syndrome or fibrilin gene mutations. Other omissions include Stickler syndrome and the other varied syndromes associated with mutations in the collagen genes.

I strongly recommend this volume to all trainees and clinicians in medical genetics who care for pregnant women or children with malformations. Others who would appreciate this book include dentists, craniofacial surgeons, maternal-fetal medicine specialists, and students and basic scientists in the fields of embryology and development. I am pleased to place it on my shelf next to Warkany's *Congenital Malformations* and the numerous embryology and development texts alongside it. I expect to reread this book regularly.

JULIE ANN NEIDICH
Biochemical Genetics and Cytogenetics
Quest Diagnostics Nichols Institute
San Juan Capistrano, CA

0002-9297/2005/7602-0025$15.00

Am. J. Hum. Genet. 76:368–369, 2005

The Genetics of Renal Disease. Edited by Frances Flinter, Eamonn Maher, and Anand Saggar-Malik. Oxford: Oxford University Press, 2003. Pp. 568. $198.50.

The Genetics of Renal Disease is an up-to-date and comprehensive review of the rapidly evolving area of genetics and nephrology. The format is that of a multiauthor book, with a total of 36 authors contributing to the 24 chapters. Most of the chapters concentrate on a specific disease that has significant phenotypic manifestations in the kidney. In addition to monogenic disorders, dysmorphic and malformation syndromes (including chromosomal disorders) are described, and the role of genetic factors predisposing to renal cancer is discussed. Also, there are more-general chapters that introduce clinical genetics, renal function and disease management, and renal development. The prospects of gene therapy for renal disease and kidney cancer are also discussed.

The three editors are from the United Kingdom and are recognized experts in different areas of renal genetic disease. Likewise, the majority of the authors are from the United Kingdom, although the author list is supplemented by many representatives from elsewhere in Europe, the United States, and Australia; each author brings expertise in a particular aspect of genetic kidney diseases. The comprehensive nature of the entire book's contents means that it will be invaluable to a wide range of individuals—such as medical and graduate students, fellows, specialized doctors, and basic researchers interested in this area. I imagine that this book would be a particularly useful reference for medical geneticists and pediatric or adult nephrologists treating a patient with a family history of an unusual renal disease.

The level of detail and the length of each chapter are relatively consistent throughout the book, although some of the more general topics and the most common diseases have required a longer description. Most chapters are more than adequately referenced, and some provide a very detailed bibliography of the subject area. The style and content varies somewhat between chapters, reflecting the individual styles or preferences of the authors, but there is a general format for the disease-related chapters. This format consists of a clinical introduction to the disease, a discussion of the pathology, the details of the genetics and molecular biology, and a description of the relevance of this information to diagnostics. The specific diseases that are covered in detail (author names in parentheses) are Alport syndrome (Flinter); autosomal dominant (Saggar-Malik and Somlo) and autosomal recessive (Guay-Woodford) polycystic kidney disease (PKD); other cystic diseases (Saggar-Malik); congenital nephrotic syndrome and focal segmental glomerular sclerosis (discussed under the strangely titled category "primary hereditary nephropathies") (Tryggvason); metabolic disorders (Town and van't Hoff); stone-forming diseases (Strazzullo and Vuotto); tubular transport disorders (Geller, Devonald, and Karet); tuber-

ous sclerosis (Weber and Mueller); neurofibromatosis (Huson and Canham); von Hippel-Lindau disease (Maher); and Bardet-Biedl and Alström syndromes (Beals, Parfrey, and Katsanis).

The enduring strength of a book like this, which covers a rapidly developing area of biomedicine, is the clinical descriptions of the disorders. These descriptions, on the whole, are sufficiently detailed to allow accurate diagnoses of the diseases and, in many cases, are well illustrated, including color plates in the center of the book that show particular clinical, radiological, and histological features of the disorders. The genetics of the disorders are clearly outlined, and those outlines will be a useful guide for the nonexpert in the field. The gene descriptions for the disorders (for which such data are known) will also serve as a well-referenced reservoir of information that should be useful for years to come. Aspects of the book that may be less enduring—because of rapid development in this area of research—are discussions of the functions of gene products and the molecular biology for genes that have not yet been identified. The authors, to their credit, have generally made their chapters as current as possible, with several references from 2003 and recent reviews cited.

Nevertheless, for some areas in which particularly rapid research progress has been made recently, the data already seem somewhat dated. This is evident for the diseases of nephronophthisis and Bardet-Biedl syndrome, for which several new genes have been identified within the past year or so and for which additional data on the localization and possible function of the encoded proteins have become available. Research on autosomal dominant PKD (ADPKD) has also developed rapidly, and, in the book, there is only a brief mention of the possible role of primary cilia in ADPKD and other forms of cystic disease. This area of inquiry is now a central aspect of PKD research and offers the prospect of a common feature linking many forms of cystic disease. Despite these inevitable problems in trying to be current in a rapidly developing area, the book remains an excellent review of these disorders. Overall, this collection of chapters comprehensively describes genetic renal diseases and will be a very useful companion for clinicians and scientists at all levels of training who are interested in this discipline.

PETER C. HARRIS

Division of Nephrology
Mayo Clinic College of Medicine
Rochester, MN

0002-9297/2005/7602-0026$15.00