PostScript

BOOK REVIEWS

Inborn errors of development: the molecular basis of clinical disorders of morphogenesis

Edited by C J Epstein, R P Erickson, A Wynshaw-Boris. Oxford: Oxford University Press, 2003, £150, pp 1082. ISBN 0-19-514502-X (hardback).

The third edition of David Smith's outstanding textbook, *Recognizable patterns of human malformation*, was published in 1982. It consisted of 652 pages with detailed descriptions of over 250 malformation syndromes. It was, and remains, an excellent book. Yet it did not contain a single mention of molecular pathogenesis or an underlying mutational spectrum.

How times have changed. Now, thanks to extraordinary developments in molecular biology, it is the exception rather than the rule to encounter a syndrome for which there is not an available molecular handle. The diverse fields of molecular genetics and clinical dysmorphology have pooled their intellectual responses to burrow away at the mysteries of human development. Their triumphant emergence is elegantly encapsulated in this wonderful book.

The title is clearly derived from Garrod's original concept of ''inborn errors of metabolism'' and it is probably not too fanciful to surmise that the next edition will come with a pullout wall chart linking all the known molecular developmental cascades in the sort of multidirectional interrelated nightmarish diagram that is much beloved of biochemists. For the moment readers will have to suffice with accounts of individual pathways and gene families. Not that they should feel deprived, for this is a very impressive review of the state of the art.

The editors have divided the 108 chapters into four main sections covering patterns of development, defined developmental pathways, gene families not yet in pathways, and regulatory processes. Each chapter is roughly equally divided between molecular biology and ''syndromology'', with valiant attempts to make plausible links between cause and effect. Inevitably some chapters score more highly than others but I would be hard pressed to find fault with any of the many chapters consulted during a busy six weeks of clinical practice. However it was a little surprising to find that some topical conditions such as Seckel's, Sotos's and Van der Woude's syndromes have not been included, and a few more illustrations would not go amiss. One particular chapter, which should perhaps remain anonymous, contains two solid pages of amino acid sequences without a single diagram or illustration to enlighten the reader.

Overall it is difficult to see how this book can be faulted, representing as it does the collective wisdom of three erudite editors, 188 distinguished authors, and a generation of outstanding scientists and clinicians. Together with the latest edition of Gorlin's *Syndromes of the head and neck*, this provides the definitive guide to molecular and clinical dysmorphology. It's not cheap but no genetics library worthy of the name can afford to be without it.

I D Young

The genetics of renal disease

Edited by F Flinter, E R Maher, A Saggar-Malik. Oxford: Oxford University Press, 2003, £120.00 (hardback), pp 568. ISBN 0192631462

''The kidney inspires awe in all those who attempt to understand its functions.'' This first sentence of chapter 2 of *The genetics of renal disease* perfectly captures the spirit of this book. Inherited renal disease is one of the most diverse, fascinating, and quickest growing areas of human genetics. Functional analyses of the genes associated with renal disease are leading to connections with many other scientific fields. For example, as is beautifully illustrated in colour plate 5 in this book, proteins now known to be directly involved in genetic renal disease are linked to the actin cytoskeleton, the endothelium, and basic cellular processes studied by laboratories not necessarily involved in renal disease.

The genetics of renal disease contains everything you might expect based on its title, and also some surprises. In the category of things you would expect are chapters devoted to each of the major genetic renal diseases, including Alport's syndrome, cystinosis, autosomal dominant polycystic kidney disease, autosomal recessive polycystic kidney disease, tuberous sclerosis complex, and von Hippel Lindau disease. An important feature is the broad scope of each chapter, most of which encompass aetiology and pathophysiology as well as clinical management. For example, the chapter on autosomal dominant polycystic kidney disease by Saggar-Malik and Somlo includes animal models of polycystic kidney disease, functional analyses of the PKD1 and PKD2 genes and proteins, mutation detection, and a flow diagram for the clinical evaluation of suspected intracranial aneurysm in autosomal dominant polycystic kidney disease. The chapter on autosomal recessive polycystic kidney disease by Guay-Woodford discusses animal and cell culture models, prenatal diagnosis, and clinical management. The chapter on von Hippel Lindau disease by Maher discusses the function of the von Hippel Lindau disease protein, clinical and genetic diagnostic strategies, and the management of renal and extra-renal manifestations.

What are the surprises? The first surprise, and a highlight of the book, is the third chapter, which is by Paul Winyard, and is titled ''Renal development''. This provides a comprehensive yet readable summary of a complex field with relevance to the pathogenesis, and perhaps eventually the therapy, of many renal diseases. The first and second chapters are introductions to clinical genetics and renal physiology, respectively. These will be helpful to non-specialists.

A second surprise is the very complete chapter, written by Gough and Vile, on gene therapy for renal cancer, which provides a potential framework for gene therapy for other forms of renal disease. Perhaps future books of similar scope will be able to include chapters devoted to targeted small molecule therapies and renal regenerative therapy.

A third surprise is the inclusion of genetically complex renal diseases such as glomerulonephritis and membranous nephropathy, and genetically complex systemic disorders affecting the kidney, including hypertension, diabetes, and systemic lupus erythematosus. The genetic data underlying these diverse diseases are elegantly summarised in a chapter by Powis. Elucidating the genetic basis of these disorders will be a major challenge of the next phase of renal genetics research.

In summary, this comprehensive yet accessible text links genetics, pathogenesis, and clinical management. The reader can rapidly locate a single piece of information using the index, efficiently review a topic by reading a few paragraphs, or acquire a comprehensive introduction to a disease by reading an entire chapter. *The genetics of renal disease* will have broad appeal to paediatric and adult nephrologists, clinical and laboratory based geneticists, and basic scientists, highlighting the increasingly interdisciplinary nature of renal disease research.

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CORRECTION

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Two authors, M Halme and A Räisänen-Sokolowski, were inadvertently omitted in the paper by Harrison *et al*, published in December 2003 (*J Med Genet* 2003;**40**:865–71). The correct author sequence should therefore be: R E Harrison, J A Flanagan, M Sankelo, S A Abdalla, J Rowell, R D Machado, C G Elliott, I M Robbins, H Olschewski, V McLaughlin, E Gruenig, F Kermeen, M Halme, A Räisänen-Sokolowski, T Laitinen, N W Morrell, R C Trembath. The error is much regretted.