PostScript

BOOK REVIEWS

Genetics for Haematologists: The Molecular Genetic Basis of Haematological Disorders

Wadie F Bahou. ReMEDICA Genetics for . . . Series. Series Editor, Eli Hatchwell. (Pp 142; £25.) London: ReMEDICA Publishing. 2000. ISBN 1 901346 11 0.

This book is a relatively small monograph of 130 pages, which is part of a series published by ReMEDICA, each monograph aimed at being a succinct summary of genetic disorders seen by a relevant medical speciality. The series aims to bring non-genetic specialists up to date both in relation to genetics in general and their own speciality. The book is effectively in two parts, the first being a list of 32 different haematological disorders, and the second being just over 30 pages of basic molecular (but not clinical) genetics, with an attached glossary. The second part is presumably common to each book in the "ReMEDICA Genetics for . . ." series. The listings for each disease are standardised under a set of headings which include the disease MIM number, gene locus, mutational spectrum, age of onset, epidemiology, inheritance, clinical features and diagnosis, special features, and gene size.

Given the need for brevity to produce a small monograph, a great deal of information is packed into each two or three page entry. There is often considerable discussion on the biological mechanisms of disease and only a brief mention of clinical details. The discussions on the nature of disease mutations are very useful, and give broad indications as to whether molecular genetics is easily applicable for the particular disease in question.

There is no weighting as to the clinical significance of disease, for example, von Willebrand's disease and the haemophilias get roughly as much detail as other far rarer disorders. Controversies over thrombotic predisposition alleles such as factor V Leiden are only briefly alluded to, and could give the impression that the issues over factor V Leiden are cut and dried.

It is also not clear why some haematological disorders are included or excluded in the book. Paroxysmal nocturnal haemoglobinuria and neonatal alloimmune thrombocytopenia are both included, although neither is a germline disease, one being a somatic mutation, and the other an autoimmune disease targeted against specific antigens. However, no mention is made of the enormous genetic contribution at a diagnostic service level to the management of leukaemias from conventional and molecular cytogenetics, and more recently from molecular genetics. The title of the book is somewhat misleading, as a haematologist might expect more on the genetic aspects of leukaemia, one of the major diseases in haematology. The first thing an undergraduate might recall of genetics and haematology is the Philadelphia chromosome, which doesn't actually get a mention.

There is a noticeable absence of illustrations, both in the disease specific section and the general genetic section of the book. Most basic genetics concepts are better explained by good illustrations, which would have added much to the book. Patterns of inheritance are not explained and the role of a clinical genetic service in the management of genetic disease is not mentioned.

I am reluctant to recommend this book to geneticists, as much of the information on any specific haematological disease is already available elsewhere. For haematologists, the book has much useful information in a snapshot form. However, for dealing with a patient with one of the diseases mentioned, a haematologist will need much more detail than is presented in the book.

Andrew J Green

Genetics for Dermatologists: The Molecular Genetic Basis of Dermatological Disorders

Sherri J Bale. (£25, $40.) London: ReMEDICA Publishing. 2000. ISBN 0 901346 10 2

Published by ReMEDICA of London in 2000, this is a short guide to the molecular genetic basis of dermatological disorders. The book is one of a similar series covering cardiology, haematology, and dermatology, with others planned for endocrinology, oncology, ophthalmology, and rheumatology.

Printed mostly in double page format, the book is separated into seven subsections, covering disorders of cornification, especially disorders of keratin and related epidermal structural proteins, such as the demogleins, loricrin, etc, most of which Dr Bale has researched. Other sections include various cancer associated disorders, bullous, pigmentary, epidermal appendageal, dermal disorders, such as Ehlers-Danlos syndrome, and mixed metabolic disorders, such as the porphyrias, premature ageing disorders, etc. A minor grouse is the author's impression that genetic dermatology was only born in 1991. It was certainly very much alive and kicking in R S Wells's group almost 30 years earlier, although admittedly recombinant DNA did not materialise in the field until the 1980s. Long before that, however, various of the chosen disorders had been illuminated by very sophisticated protein separation, enzyme assay, or electron microscopical analysis, as well as traditional pedigree analysis.

Individual sections are organised by eponym, MIM number, clinical features, age of onset, inheritance, chromosomal location, genes responsible, mutational spectrum, diagnosis, and counselling issues. These are generally mutually exclusive, but sometimes overlap significantly. Eighty-six genetic diseases are included, of which 29 are illustrated. Of these, only occasional pictures, such as Hermansky-Pudiak syndrome, ectrodactyly, and porphyria cutanea tarda, were of the standard expected. The majority were generally too small, indistinct, or non-specific to be diagnostically useful. Examples of the latter group include epidermolytic and non-epidermolytic palmoplantar keratoderma, which were too similar to separate, although histologically they would have been. Similarly, the illustrations of EB simplex and severe dystrophic EB do not clearly show the tense superficial bullae of the one, nor the severe crippling and distortion of the other. Similarly, the vascular EDS IV illustration does not show the crucial epidermal atrophy, acral features of the disease.

Competitors of this book include McKusick's Catalogues (still the gold standard text) and sections within large textbooks (such as the 6th edition of Rook's *Textbook of Dermatology*, which is just as informative, better illustrated, and more logically organised by chromosomal location). Both of these two volumes are very bulky, unlike the alphabetically organised *Illustrated Dictionary of Dermatologic Syndromes*, which is famously succinct, portable, and very informative, although now over seven years old.

So much for the weaknesses. The strengths of *Genetics for Dermatologists* include topicality, portability, a reasonable price, and up to date information on gene structure and function. It would be a useful addition to the shelves of aspiring dermatological geneticists and general geneticists with interests in dermatology. At £25 ($40) it is slightly expensive for its 230 pages, but still reasonable value for money.

Priority considerations for the 2nd edition would include larger, clearer, and more comprehensive illustrations, a loose leaf format with the option for continued updating, more details of gene structure and comparative properties of non-allelic variability (as in neurofibromatosis and tuberous sclerosis), and genotype-phenotype correlations of individual genes such as collagen type VII and EB dystrophica. Similarly, succinct comparative tables of the keratin pairings would be very useful. Last but not least, the names and addresses of key UK, USA, and European gene and/or protein electron microscopists would be extremely useful where pertinent to disorders of keratin, pigmentation, matrix proteins, enzyme assays, etc. No doubt this would greatly facilitate the task of those non-experts to whom the world of www and general internet access is still foreign, puzzling, or unavailable.

F M Pope

CORRECTION

In the August 2001 issue of the Journal (*J Med Genet* 2001;**38**:536-40), in the Letter by Horvath *et al* on "High frequency of the *ApoB*-100 R3500Q mutation in Bulgarian hypercholesterolaemic subjects", the estimated prevalences of the R3500Q mutation in the Czech Republic and Slovakia, shown in fig 2, have been transposed and the correct prevalences should be "no data available" for Slovakia and 1:250-1:350 for the Czech Republic. The authors apologise for this error.