Book reviews

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Endocrine Genes: Analytical Methods, Experimental Approaches, and Selected Systems

Edited by Yun-Fai Lau. (Pp 224; £30.00.) New York: Oxford University Press. 1988.

The book is the result of a series of research seminars held in the University of California in the autumn of 1984. The authors have clearly tried to update their presentations but, from the state of the methodology and the reference lists, it is quite clear that the book is already considerably dated. What is not in doubt, however, is the fact that the contributors are from groups of excellence with established reputations who have very much been at the centre of developments in molecular endocrinology. Despite the fact that it is a multi-author production, the standard of the presentations is remarkably uniform and high and I certainly enjoyed reading this book and learned from it. It is difficult to know what its ideal readership would be, but there is no doubt that any endocrinologists aspiring to the application of recombinant DNA technology to problems in endocrinology would be well advised to peruse this manuscript. The book essentially seeks to highlight developments in technology and provide examples of their application and it does this well. Clearly from the size of it, the information that it contains relating to methodology is somewhat limited, but the excellent referencing will ensure that readers can track down more specific information, if they require it. My initial criticism that the work was somewhat dated because of the rapid developments in the field in no way diminishes the value of this book as a starting source of information in molecular endocrinology.

A M MCGREGOR

Mendelian Inheritance in Man. Catalogs of Autosomal Dominant, Autosomal Recessive, and X-linked Phenotypes

8th edition. By Victor A McKusick. (£47.00.) Baltimore: Johns Hopkins University Press. 1988.

It has not been easy to review the 8th edition of 'McKusick' (the chocolate brown one) because whenever I decided to look at it in detail I found that someone in the department had borrowed it. The

fact that it has been in constant use since it landed on my desk a month ago is perhaps the best recommendation of all. Quite simply all clinicians, not just those involved in genetic disorders, should have access to this book through their medical library or in their own department. Some medical geneticists will want their own personal copy. Those clinicians who claim that their practice does not involve disorders inherited in a Mendelian fashion are probably wrong!

Mendelian Inheritance in Man (or MIM, as Dr McKusick abbreviates it) is an ideal way of getting into the body of published reports for those clinicians who rarely deal with a particular Mendelian disorder. The value of MIM as a starting point for a search of publications is, of course, well known to the majority of readers of this Journal, and yet I can recall many times when I have opted to search elsewhere, perhaps in a more specialised book, only to find in the end that it would have been best to have gone straight to MIM.

Since the 7th edition (1986) the total number of entries has increased by 437 to 4344, of which 2136 are in the provisional category, representing loci that are not fully identified or validated and therefore do not receive an asterisk. The total number of different references cited is now 31 278 (with 48 551 authors). There are also some new features in the appendices to the Foreword. The section on the Human Gene Map is greatly improved and much more user friendly. I had found earlier human gene maps, particularly the section dealing with disorders for which the mutation had been mapped to a specific site, difficult to use because of the lack of an alphabetical listing of the disorders. This has now been put right. The gene symbol list, which lists each locus by its approved symbol in alphabetical order, is also arranged better than before. Sometimes the symbol is not so obvious as to allow one to find a particular phenotype from the list itself, but one can always use the main index and get the symbol from the entry in the book. This also provides the reference(s) on which the map position is based.

The section dealing with molecular defects in Mendelian disorders has been simplified. This has evolved out of various different listings over the last few editions, and now has a single list of about 400 disorders in alphabetical order. Included for the first time is a column for the intragenic lesion classified

into Point mutations (seven varieties) and Length mutations (four varieties).

The incorporation of all this new information is very impressive but it is creating a problem that Dr McKusick recognises but only partially solves. Despite the book's subtitle 'Catalogs of autosomal dominant, autosomal recessive and X-linked *phenotypes*', the book is moving towards being a 'listing of loci, not phenotypes. MIM is an encyclopedia of gene loci'. The eventual aim is to have a single asterisked entry for any properly characterised gene locus, even though different mutations at this locus may give clinically distinct disorders. This is clearly a sensible move since in the end the most secure classification of Mendelian disorders must be by the gene locus involved. This makes sense both in understanding the pathogenesis and in the use of diagnostic tests based on DNA analysis. Once two disorders are established as defects at the same locus, such as the Duchenne and Becker types of muscular dystrophy, they are to be included in a single asterisked entry. The 7th edition gave Duchenne (31020) and Becker (31010) muscular dystrophy separate asterisked entries, but in the latest edition there is a single asterisked entry 'muscular dystrophy, pseudohypertrophic progressive, Duchenne and Becker types' (31020). The number 31010 disappears.

However, it has to be recognised that clinicians have, and will continue to have, good reason to distinguish Duchenne and Becker muscular dystrophy in the same way that they must recognise sickle cell anaemia and β thalassaemia major as distinct disorders, even though they are both mutations at the β globin locus. Sickle cell anaemia (or trait) or haemoglobin S no longer appear in the index, having been removed after the 6th edition in 1983. This is unfortunate. It can be argued that only the extremely ill informed will not think to look it up under Hemoglobin- β locus, but when Duchenne or Becker muscular dystrophy are eventually replaced solely by muscular dystrophy-dystrophin locus, and Stickler's syndrome solely by Arthro-ophthalmopathy, hereditary progressive-COL2A1, and so on, it will be disheartening for interested clinicians. They should be led to the correct locus and agreed clinicopathological designation by abundant cross referencing of the commonly used disease names in the index. I am sure this problem is solved by use of the computerised online version, OMIM, but that is no reason to overlook the difficulties in the 'hard copy' editions.

I recognise the difficulties of multiple names for the same condition and in drawing the line on which mutations can be regarded as causing a disease (should haemoglobin E be indexed or lumped under

Hemoglobin variants?), but that is no reason not to draw a line. If anyone is up to this difficult task it is Dr McKusick, who moves so ably between clinical medicine and basic human genetics.

Reviewing the 8th edition has given me reason to become reacquainted with the Foreword. These 12 pages should be required reading for all clinical geneticists in training. There is only a small addition to the text for the 8th edition; namely a comment on the elucidation of the mutations in the transthyretin (thyroxine binding prealbumin) gene (17630) that underlie the several forms of hereditary amyloid polyneuropathy: the Andrade, or Portuguese type (10480); the Rukavina, or Indiana type (10490); the Cardiac, or Danish type (10500); and others. These entries illustrate the move to a single *asterisked* entry per locus. The Andrade or Portuguese type entered as Amyloidosis I gets the asterisk. The other types and the transthyretin gene retain a numbered entry, but no longer have an asterisk. I cannot quite see why the above three amyloid polyneuropathies are not now all under a single entry, but no doubt this will come. It is important for readers to appreciate that there are now two reasons for an entry not having an asterisk; either there is still doubt as to whether the phenotype can be the result of a mutation transmitted in a Mendelian fashion, or that entry refers to a locus that already has an asterisked entry. As discussed in the Foreword there are considerable constraints on the organisation of the information imposed by evolution of the 8th edition from previous catalogues going back to 1966. I can see a time when ready access to all the information will only be easy using the online version. However, the book will, I suspect, be the main outlet for 'McKusick' for many years to come. It will be increasingly used by a greater variety of clinicians as they come to recognise that genetics is an integral part of modern medicine. I just hope our copy survives; the binding does not look as robust as in previous editions and it is already looking 'well used'.

MARCUS PEMBREY

The Cytogenetics of Mammalian Autosomal Rearrangements

Edited by Art Daniel. (Pp 938; \$295.00.) New York: Alan R Liss. 1988.

Autosomal rearrangements are of considerable importance in human and animal populations. This book, which, for the most part, consists of a series of review papers from acknowledged experts, is an attempt to provide an overview of current knowledge