

**Neurology of Hereditary Metabolic Disorders of Children.** G Lyon, R D Adams, E H Kolodny. (Pp 379; \$75.00.) New York: McGraw-Hill. 1996. ISBN 0-07-000389-0.

There is a need for a book on hereditary metabolic disease of childhood which is user friendly, not too large to pick up, not in two volumes, and written primarily for the clinician. This book fulfils all of these criteria. It has even been printed on acid free paper (could it be more user friendly?), and although the book will not fit neatly into your pocket (it is fairly comprehensive) it will not conuss you either when it falls off the shelf.

The authors, who are well respected clinicians in the field, have taken the unusual stance in deciding not to describe disorders according to their chemistry. The chapters are not entitled "organic acid disorders" or those of "purine and pyrimidine defects" but are subdivided according to age of onset and this is just how a clinician would begin to think. The first chapter, therefore, deals with disorders presenting in the neonatal period, and then it proceeds through early infantile onset, late infantile, into childhood and adolescence. This does necessitate some degree of repetition. The discussion of Krabbe's disease, for instance, which might present in any of those age groups, is dealt with in various chapters, but this does not seem to matter as the infantile onset disease is so different from that appearing in childhood. Laboratory tests, and the latest genetic linkage information, are well covered. The primary defect in Smith-Lemli-Opitz syndrome is mentioned and prenatal diagnosis has not been neglected.

What is particularly pleasing is that the authors bring their own clinical practice with sentences like "to an experienced neurologist intention myoclonus ... always conveys the idea of hereditary metabolic disease", and the text is full of tips. They are not frightened to say "in our experience there are five major troublesome areas", and these are listed in point form and discussed.

Another outstanding feature is the line drawings indicating the clinical course and life profile of well known diseases showing when different clinical features appear in the course of deterioration. There is also a helpful perspective in that the authors are prepared to be fairly dogmatic. They would state "in some metabolic encephalopathies seizures are obligatory signs", and the reader knows they are in the hands of experts and would take heed. This book should be read, and I mean read, by all paediatric neurologists of whatever age. This reviewer remembers many years ago, when a senior registrar in neurology in Cape Town, being visited, and suitably put in his place by the now, I take it, very senior author of this book (RDA), and being astounded by his erudition. He and his co-authors are still imparting erudition with enthusiasm.

MICHAEL BARAITSER

**An Introduction to Recombinant DNA in Medicine.** 2nd edition. A E H Emery, S Malcolm. (Pp 206; £14.95.) Chichester: John Wiley. 1995. ISBN 0-471-93984-6.

The authors' stated aim is to "provide an introduction to the subject of recombinant DNA technology for those whose interests are perhaps more medically orientated ... (giving) a simple outline of the general principles of DNA technology and some of the ways in which it can be applied".

In writing this illustrative rather than comprehensive text, they have been broadly successful in fulfilling this aim. To distil the essence of a subject is more difficult than merely recording every last detail.

The first part of the book summarises the structure and function of DNA, DNA technology, and gene mapping, structure, and function. Although some sections are astonishingly brief (for example, that on protein translation), the text is clearly written and conveys concepts and strategies rather than being overloaded with nomenclature. However, some of the diagrams are unclear; for example, the one to explain Sanger sequencing is confusing. In addition, the majority of the first chapter could be dropped without loss; most of the information is duplicated elsewhere.

The next two chapters deal with the molecular pathology of disease. Firstly, the principles of single gene disorders are discussed, with particular focus on the haemoglobinopathies, followed in the subsequent chapter with examples of the causation of common diseases such as diabetes. A highlight of the book is the section on cancer genetics, an excellent summary of the field.

Prevention of genetic disease through prenatal diagnosis is covered well and uses Duchenne muscular dystrophy to illustrate the range of methods available. The last major chapter alludes to some of the treatment options, again, a very neat summary.

Overall, this short book is a good starting point for someone who wishes to introduce themselves to molecular medicine, and it should entice them to read further.

FIONA NORWOOD

**Human Molecular Genetics.** Editor Kenneth W Adolph. (Pp 500; \$85.00.) New York: Academic Press. 1996. ISBN 0-12-044310-4.

There are now a large number of books available which describe the theory and techniques of molecular genetics, many of which include protocols for the experimental techniques. This book is the eighth volume of a series of such books describing methods in

molecular genetics, and deals in particular with human molecular genetics. It is a comprehensive book dealing with many newer techniques and covers a wide range of subjects from gene mapping and mutation detection to transcription, RNA editing, signal transduction, and the use of the mouse as a model system. It is aimed at providing practical experimental advice for experienced scientists and achieves this well.

This is a useful addition to the body of publications and is well thought out. There are 26 chapters separated into seven sections: mutation detection in human genes; gene mapping, cloning, and sequencing; transcription, promoters, transcription factors, mRNA, RNA editing, ribozymes, antisense RNA; genome recombination, amplification; receptors, signal transduction; the mouse as a model system for human molecular genetics. Each chapter covers a different technique and includes a brief explanation and detailed protocols. The techniques described are dealt with very well and in enough detail to allow the reader to attempt the techniques based on the information given. Many topics are covered but there are, however, some omissions. In the section entitled gene mapping, cloning, and sequencing, for example, there are chapters describing direct cDNA selection, subtraction cDNA cloning, and fluorescent differential display but no mention is made of exon trapping.

As an introduction to the techniques some of the chapters are excellent and provide the background information necessary to understand and interpret results, but the book covers such a wide range of techniques that it is unlikely that one person would want to read every chapter in detail. The format is rather dry and the book would not be a good teaching aid, but for the experienced scientist would be a good reference text. I am not sure I would rush out to buy a copy but would find it helpful to refer to occasionally and would be glad to find a copy in the genetics department library.

DOROTHY TRUMP

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## NOTICE

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### 38th Annual Short Course in Medical and Experimental Mammalian Genetics

This course will be held on 14-25 July 1997 at The Jackson Laboratory in Bar Harbor, Maine, USA. For further information contact: 1997 Courses and Meetings, The Jackson Laboratory, tel: (207) 288-6262; fax: (207) 288-6080; Email: education@aretha.jax.org; URL: <http://www.jax.org>.