Book reviews

The Principles of Human Biochemical Genetics

This is the third edition of the standard book on human biochemical genetics published in the English speaking world. Though entitled ‘Principles’ it is sufficiently comprehensive in coverage of most of the important and interesting facts of the subject to serve as a textbook. In his prefaces to the three editions Harris has never made it clear at whom the book is aimed. Perhaps there is no need to do this, for there are items to please and instruct even those with the most casual interest in biochemical genetics, while for the professional the book is a goldmine.

Surprisingly, in view of the 5 years which have elapsed since the previous edition, the size has increased by less than 30%. The format is essentially unchanged. ‘Inborn Errors of Metabolism’ has been expanded and divided into two parts. ‘Gene Mutations Affecting Rates of Protein Synthesis’ has been dropped, since α-thalassaemia and HPFH can be satisfactorily dealt with under deletions. The HLA system has at last been recognised, having been ignored in earlier editions, but coagulation disorders are still not seen as part of biochemical genetics. I am distressed that a former Galton professor should allow his spelling to be Americanised by his Dutch publishers.

What about the ‘new genetics’? Here Harris must be criticised. He devotes only three pages to restriction endonuclease mapping and five to polymorphisms at the DNA level in a total of 440 pages of text. For a book dated June 1980 this is hardly adequate. Exclusion of DNA from the subject index is another peculiarity. Biochemical genetics is going to be turned on its head by the powerful new tools which recombinant DNA technology has provided. Traditional biochemical geneticists may regret this and wish to continue working with the starch gels which have been so productive in the past. But there really is no alternative but to turn away from protein variants and grapple with the complexities of DNA biochemistry.

D J H Brock

Practical Genetic Counselling

In my enforced inactivity during a recent holiday, Harper’s book demonstrated its excellence. It was small enough to have been taken along in the first place, and by its pleasant style it diverted the mind from the somatic consequences of inexpert skiing. The essences of this book are its portability and completeness. It is an indispensable aide memoire for anybody concerned with genetic problems in medicine. Certainly paediatricians, obstetricians, and many others will find it easy to read and an unfailling source of rapid reference. I have no doubt that most clinical geneticists will also keep a covert copy in their briefcases, especially on peripatetic visits. Perhaps it is only genetic paranoia that makes one feel that one ought to have total recall, but Peter Harper’s book certainly provides enough to get by in difficult situations far from one’s library. As a crib it is splendid, but it is more than that. The book describes in a readable and comprehensive way the basics of medical genetics taken to an appropriate level of complexity, falling just short of the point at which most clinicians will become confused. I strongly suspect that many people will understand for the first time the use of Bayes’s method when reading this book. Its introduction to the history of the subject leads naturally into the present scope and the organisation of genetic clinics and one is presented with an irrefutable case for the importance of properly trained clinicians in the field of medical genetics.

The sections concerned with diseases of the different organ systems are extremely helpful. It is intended that these should be for the non-specialist (inherited disorders of neurology for non-neurologists, etc.), but here again, most clinical specialists will be grateful for a readily accessible aide memoire to remind them of the inheritance of a particular disorder (whether in their specialty or not). The references provide ready access to the literature and are well chosen, but should be used in conjunction with other sources for the selection of primary material.

It would be cavalier to list the relatively small number of errors and omissions and I encourage people who spot peccadilloes to write to Peter Harper so as to encourage him to produce regular
up-dated versions. This book is an indispensable working guide to medical genetics as practised in the clinic rather than the lecture theatre.

**Rodney Harris**

**A Handbook of Clinical Genetics**

By J S Fitzsimmons with the assistance of E M Fitzsimmons. (Pp vii +159; figures + tables. £5·15.) London: Heinemann. 1980.

It is stated in the preface to this book that it was originally planned for nurses. It more than fulfils the need of nurses, both in training and when working in paediatric or obstetric departments, to understand the principles of elementary genetics and particularly their application in the genetic counselling clinic, although some mention of the genetics of blood groups would have been useful.

Good communication is one of the essentials of genetic counselling; the standard of communication in the script and also in the illustrations, both serious and amusing, is uniformly high throughout the book, whether the author is dealing with the genetic code, inheritance patterns, prenatal diagnosis of genetic disease, or ethical issues, to mention but a few of the topics covered. Perhaps it is because so many aspects of the subject are discussed that what is covered in breadth is lacking in depth, but this seems inevitable in a book of this size.

Even so, many doctors, as the author again suggests in his preface, who have not received or have forgotten any basic training in biological genetics will find it a useful and readable handbook. Furthermore, the medical students of today, who should be aware of genetics at a more advanced level than the authors would claim to describe, will doubtless benefit from this introduction to the practical application of their knowledge to the problems they will meet in practice.

**Mary Vowles**

**Genetic Metabolic Diseases. Early Diagnosis and Prenatal Analysis**


Our understanding of genetic metabolic diseases has advanced rapidly during the past 20 years, particularly with regard to the molecular basis of their pathological manifestations. A major practical benefit is that biochemical methods are now available for the accurate diagnosis of a large number of inherited disorders. Professor Galjaard's book deals authoritatively and comprehensively with this information as it relates to the 60 conditions for which prenatal diagnosis is currently possible.

The first chapter provides information on the incidences and recurrence risks of monogenic defects, chromosomal anomalies, and congenital malformations, and ends with an introduction to the biochemical aspects of early diagnosis and prenatal detection. The second chapter, taking up half of the book, begins with a description of the early clinical and developmental abnormalities associated with metabolic disease. It then continues with individual sections dealing with specific disorders in the metabolism of carbohydrates, mucopolysaccharides, muco-lipids, lipids, amino-acids, nucleic acids, and other metabolites. The sections are exhaustively referenced with key information on differential diagnosis, clinical features, pathology, and specific biochemistry, including molecular diagnosis. These are scholarly, critical accounts and probably the best to be found by anybody wanting a short yet comprehensive reference to a particular disease. There is some inevitable repetition between sub-sections and perhaps understandably in a work of this kind, a sprinkling of obvious textual errors.

Chapter 3 describes the methodology of prenatal diagnosis, including amniocentesis, AFP measurement, cell culture, and chromosome analysis. It ends with a good account of the microbiochemical techniques pioneered in the author's laboratory. There are some particularly good photographic illustrations. Chapter 4 describes current practical experience of prenatal diagnosis, each disorder being treated individually in metabolic sub-sections, as for chapter 2. Again, this has led to some repetition. Also, the experimental detail given for individual cases might be felt to be excessive sometimes. Nevertheless, it is a critical assessment which stresses the problems as well as enumerating the successes.

The next chapter discusses carrier detection and prenatal diagnosis of X linked disorders, and the final chapter deals with future prospects including enzyme replacement therapy, fetoscopic sampling and DNA analysis by recombinant DNA techniques. There is a valuable practical appendix giving sources of references and essential details for 75 biochemical and related techniques.

This is an excellent book, packed with all the information of a standard reference work, while still retaining the interest to make it enjoyable reading. The book can be highly commended to all who are concerned with the genetic aspects of metabolic disorders.

**A D Patrick**