Lysosomal Storage Disease: Biochemical and Clinical Aspects

This is an excellent book which provides an up to date and comprehensive account of the lysosomal storage disorders. There are general chapters on the diagnosis, the genetics, and the treatment of these conditions with chapters on the individual groups of disorders including the sphingolipidoses, the mucopolysaccharidoses, glycoproteinoses, mucolipidoses, acid lipase deficiencies, and glycogenosis type II. The complex biochemistry is clearly presented together with sections on the clinical phenotypes, pathology, diagnosis, and the genetic aspects.

This book will be useful for all those who are concerned with patients with lysosomal storage disease, both clinically and in the laboratory. It is up to date with some references published in 1985 in the bibliography.

My only minor complaint about this book is that it is not really as useful as it might be if you do not already know the diagnosis. Thus for those who are faced with a patient who might have lysosomal storage disease, it would be quite difficult to work out which of the disorders are likely candidates if you did not already have a working knowledge of the various conditions. The simple addition of a table of presenting symptoms and signs, for example, corneal clouding, with a list of the disorders that might be responsible, would be very useful. In addition to this, the table might include the presence or absence of vacuoles in white blood cells and whether the characteristic cells are present in marrow. All this could save time for the patient and unnecessary expensive laboratory tests. Despite this, the book is a useful source of information and very readable. I can certainly recommend it.

J V Leonard

Human Genes and Diseases
Horizons in Biochemistry and Biophysics Volume 8. Edited by F Blasi. (Pp 676; £45.00.) Chichester: John Wiley. 1986.

This book consists of 16 detailed review articles or chapters, written by different authors, which present scientific and theoretical approaches to topics in molecular biology related to human genetics. The reviews are cleverly grouped together to give a broad overview of recent developments; there are five chapters on X linked genes, a selection of autosomal disorders, and finally a group of chapters dealing with oncogenes and the structure and function of insulin and epidermal growth factor receptor genes. Some chapters were easier to follow and more interesting than others which reflects the multi-author style. I was personally interested in those sections likely to have immediate clinical relevance but was also fascinated by the developments which are taking place in the field of oncology.

The presentation of the book merits comment. At first glance the variation in typescript from chapter to chapter is disconcerting but much more important are the frequent typographical errors and occasional translation difficulties. This overall effect is further compounded by the substitution of figure 1 on page 64 for figure 4 on page 72 and pages 403 and 404 are printed in the wrong order. These mistakes are distracting. The editor claims that this book will be useful for a wide spectrum of readers: experts in molecular biology, medicine and biology students, physicians, and biochemists. I feel that its appeal will be more limited than this as a prior knowledge of the vocabulary of molecular genetics is assumed. Its use will be as a book to ‘dip into’ for reviews on particular topics rather than as a book to be read from cover to cover.

O Quarrell

Planning for a Healthy Baby: A Guide to Genetic and Environmental Risks

Professor Goodman, like so many other practising clinical geneticists, believes that prospective parents deserve help to plan for a healthy baby and he has produced an attractive, lucid, and extremely informative text to meet this need. He adopts a question and answer format and in 260 pages manages to deal with 400 items on genetic and environmental risks in pregnancy, all reputedly based on his day to day clinical experience. These cover basic facts relating to teratogens and maternal illness and include descriptions of some common birth defects. There is good coverage of simple genetics with helpful explanations of modes of inheritance, chromosomal abnormalities, and common genetic disorders. The section on diagnostic and therapeutic procedures is particularly useful. The author’s brief look into the future reflects a mature concern and awareness of normal parental fears and attitudes.

Will it suit the audience to whom it is addressed? The answer has to be a qualified yes. It should be