Lysosomal Storage Disease: Biochemical and Clinical Aspects
By R W E Watts and D A Gibbs. (Pp 284; £35-00.)

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 glycosogenosis 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

Planning for a Healthy Baby: A Guide to Genetic and Environmental Risks
By Richard M Goodman. (Pp 269; £10.95. $16.95.)

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
very useful for educated, biologically aware, motivated parents, such as are more likely to be found in North America or presumably Israel. Professor Goodman’s natural habitat. Sadly it is a lot less likely to be understood by the majority of adults in the UK, where education in biology relevant to health and reproduction has been neglected for many years. The text assumes a familiarity with biological terminology and the concept of risk, which is unfortunately rarely the case with the average British parent.

Nevertheless, there are other audiences for whom it can be recommended. It should certainly be useful for the primary health care teams, practising midwives, and perhaps even some practising obstetricians, to whom many of the questions are regularly addressed in their day to day work. The direct, simple style should also guarantee the book a warm welcome in some departments of clinical genetics.

I have no real criticisms. At $16.95 it is expensive for its targetted lay audience in this country and a cheaper paperback edition would be more appropriate for the British market. There is a surprising absence of any reference to the emotional consequences of pregnancy termination for genetic reasons. This should be rectified in future editions. Some of the selected book references are more suited to a medical than a lay audience. I doubt if many parents would want to curl up with Dr McKusick’s superb catalogue for bed time reading! These are minor points; the book deserves to be widely read and I enjoyed it.

J S Fitzsimmons

Duchenne Muscular Dystrophy

It showed great foresight on the part of Alan Emery and the publishers to produce a book devoted entirely to a disease that has probably had more written about it in the 1980s than any other condition. It was a wise decision: the monograph is not just an account of what everyone knows. It gives a comprehensive and critical assessment of every aspect of Duchenne muscular dystrophy, to suit research workers, clinicians, and physiotherapists and others who are concerned with the management of muscular dystrophy.

After a brief introduction to the physiology and anatomy of muscle, there is a fascinating history of the first descriptions of Duchenne muscular dystrophy. Here we learn that the first diagnosis was made by Dr Meryon of St Thomas’s Hospital, who also reported an X linked pedigree, about 17 years before Duchenne. It is interesting later in the book to read a reprint of the obituary of Duchenne which appeared in the Lancet of 1875. Photographs of neurologists of the last century are accompanied by a modern one of P E Becker who described the more benign type of X linked muscular dystrophy during the lifetime of many patients alive today. Chapter 3 is remarkable for some valuable tables presenting the variation in the natural history of the disease, based on Professor Emery’s large series of personal cases. These are followed by a critical account of investigations used in the diagnosis of Duchenne muscular dystrophy, and in its distinction from other neuromuscular diseases. Emery discusses biochemistry and possible pathogenetic mechanisms, and later there is a long and useful chapter on management, in relation both to individual patients and to the liaison between community services and lay organisations.

The chapters on genetics, genetic counselling, and molecular biology are particularly interesting to readers of this Journal. These chapters are very clear, practical, and up to date. The approach is wide ranging and non-controversial, although Professor Emery does point out some features of Duchenne muscular dystrophy that are not fully explained. He asks, for example: Why is the disease so common? Do postzygotic mutations occur? Why are some boys mentally retarded? Why is the proportion of female carriers detected through creatine kinase estimations different from that estimated from the proportion of affected sons? Why do DNA deletions fail to correspond with the clinical picture? Why do some families have more than one new mutation for Duchenne muscular dystrophy?

This book is timely and useful; it stimulates as well as teaches. I can recommend it for all those doctors, scientists, and paramedical staff who work with muscular dystrophy, and probably some parents would also find it interesting.

Sarah Bundey

Human Genetic Diseases: A Practical Approach

The contents of the various chapters in this volume of the, now quite extensive, Practical Approaches Series covers both routine and research applications of molecular genetics as applied to human genetic disease. The first chapter by John Old is called ‘Fetal DNA analysis’, but in addition to a discussion