Book reviews

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 life time 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 local 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 diseases. The first chapter by John Old is called 'Fetal DNA analysis', but in addition to a discussion

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

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The book reviews section contains book reviews of various books on different topics. The reviews are written by different authors and cover a range of topics, from genetics to pathology.

One review is for a book on genetic disorders and the fetus, which is published by Aubrey Milunsky and edited by Michael Milunsky. The book contains practical applications of prenatal diagnosis to chromosomal disorders and is reviewed positively for its comprehensive content and utility for genetic counselors, obstetricians, and laboratory personnel.

Another review is for a book on genetic aspects of developmental pathology, edited by Enid F. Gilbert and John M. Opitz. The book contains selected papers and proceedings of a symposium held in September 1985 in Madison, Wisconsin, USA, as part of the interim symposium of the Society of Pediatric Pathology. It contains a mixture of papers, ranging from good reviews through 'oddities we have seen in our department over the last two or three years' type papers to single case reports. There is much that will be useful to the clinical geneticist or pediatric pathologist: reviews of the developmental pathology of collagen, pathological findings in 5,10-methylene tetrahydrofolate reductase deficiency, glutaric acidemia type II, neonatal haemochromatosis, hepatic involvement in hereditary renal syndromes (an excellent review), lissencephaly syndromes, and more besides. Of the personal experience papers, I would single out that by Dr Dagmar Kalousek on anatomical and chromosome anomalies in specimens of early spontaneous abortion as being extremely useful. All in all a very informative book to have on the shelves of a genetics or pathology department.

ROBIN M WINTER

Genetic Disorders and the Fetus

The first edition of Genetic Disorders and the Fetus appeared in 1979, at the end of the first decade of the practical application of prenatal diagnosis to chromosomal disorders, inborn errors of metabolism, and congenital malformations. As a comprehensive summary of the state of the art it had few peers, and it became the reference text for genetic counsellors, obstetricians, and laboratory personnel. More than half of the first edition was the personal work of Aubrey Milunsky, whose command of the literature and dedication in seeking out obscure references was one of its most impressive features.

The uses of prenatal diagnosis have expanded considerably over the intervening seven years, and this fact is recognised in the second edition, which has become an edited text with 46 contributing authors. There are new chapters on the fragile X syndrome, on the haemoglobinopathies, on connective tissue disorders, and on flow cytometry of metaphase chromosones. The section on metabolic disorders is divided into ten separate chapters, with various experts called in to give breadth and depth to the presentations. The only casualty from the first edition is the omission of the chapter on the role of infectious agents in the causation of birth defects.

The most important new developments in prenatal diagnosis are the use of chorionic villus sampling for first trimester detection of affected fetuses and the application of molecular genetic...