Book reviews

myotub is thus very complicated; however, its study in muscle culture will hopefully lead to an understanding of how dystrophin exerts such an important effect on muscle function, a theme taken up by H J Klamut et al. The animal models described by B J Cooper are also likely to be useful in understanding the pathogenesis of Duchenne muscular dystrophy, particularly the point mutation in mdx mouse which causes no clinical disease. L M Kunkel and E P Hoffman summarise the exciting work of analysing the dystrophin gene, and emphasise the curious feature of its size, which is disproportionately large for the small protein it encodes, and which is presumably the reason for its frequent mutations. D R Love et al suggest that study of the dystrophin gene of mildly affected Becker patients might lead to the construction of an in vitro engineered gene that could be suitable for gene therapy.

S V Hodgson and M Bobrow describe how carrier detection is more accurate when DNA probes are used in conjunction with creatine kinase estimation, and how prenatal diagnosis is available for the majority of female carriers: a great advance. J T den Dunnen et al report that field inversion gel electrophoresis is a rapid way of detecting rearrangements and deletions of the dystrophin gene and is useful in recognising female carriers. They find that 14% of new mutations are associated with germinal mosaicism in the mother.

There are further chapters on the management of muscular dystrophy, on myotonic dystrophy, and on the differential diagnosis of Emery-Dreifuss and other muscular dystrophies. A E Harding and I J Holt succinctly describe the intriguing findings in the clinically diverse mitochondrial myopathies. These are associated with a variety of biochemical abnormalities in mitochondrial respiratory complexes, some of which result from mutations in mitochondrial genes and others from mutations in nuclear genes. The authors were the first to describe heteroplasmy in humans, by showing that some patients with progressive external ophthalmoplegia or Kearns-Sayre syndrome had deletions of the mitochondrial genome in some, but not all, of mitochondria within individual muscle cells. Presumably this somatic mosaicism (or, more accurately, heteroplasmy) accounts for some of the clinical diversity and for the rarity of inheritance from a parent.

This book thus ranges widely over many muscle disorders and will be valuable to geneticists and neurologists, and also to scientists trying to unravel the basic molecular and biochemical defects. It is compact and well presented and I thoroughly recommend it.

SARAH BUNDEY


I enjoyed reading this short book on prenatal diagnosis edited by Whittle and Connor, which covers many aspects of the subject, starting with the general principles of counselling, some principles of screening, and many fairly detailed chapters on structural abnormalities. There is a very brief chapter on DNA diagnosis of single gene disorders, but nothing here that can help an understanding of mathematics of precise risk prediction with linked markers where crossover is a possibility.

The book contains a very large number of good ultrasound pictures. Unfortunately, as with so much writing on ultrasound, the authors work backwards from pathological diagnosis to the clinical appearances, instead of working forward from what is seen on ultrasound to the possible pathological diagnoses, giving an indication of which is most likely and an overall view of prognosis.

This is a competent book and will be of some value to people working in obstetrics. It is not sufficiently detailed for a sub-specialist trainee in the area and it is rather too detailed for a general practice readership. It would, however, be useful for an enthusiastic candidate for the MRCOG. One of the best features of the book is a list at the back of the various diseases which can be diagnosed by a chemical assay and another of the various structural malformations that have been picked up on scan.

Unfortunately, this book shares, with many other publications, an inflated view of the precise diagnostic accuracy of ultrasound. All sorts of unusual and specific conditions are claimed to be diagnosable by ultrasound. What happens in actual practice is that an unusual appearance is noted on the scan. Then, after delivery, the precise diagnosis is made available. The authors then write up the case as if the condition was precisely diagnosable antenatally.

Nevertheless, notwithstanding this rather general error, I was quite impressed by this little book.

R J LILFORD


The primary aim of this book is to help cytogenetists and clinical geneticists provide risk figures for the occurrence of chromosome abnormalities under defined circumstances, and to indicate the likely phenotypic outcome. This endpoint suggests that the text is a tedious list of facts and figures. On the contrary, the information is supplied in an informal but concise manner.

All clinical geneticists and cytogenetists are familiar with the debates that can ensue on detection of a chromosome abnormality that is not associated with a well established syndrome. Sometimes appropriate reprints are to hand: more often, a review of previous publications is instigated, records (and memories) are searched, or colleagues contacted, to determine whether or not a similar case exists. The authors of this book have carried out all of these tasks and compiled the data so that they are readily accessible and comprehensive (there are over 500 references).

There is an introduction to the subject matter in the first three chapters, ensuring that the less genetically aware readership is reminded of the basic concepts of cytogenetics, the mechanics of meiosis, and the derivation and application of risk figures. Thereafter, each chapter is devoted to a particular aspect of cytogenetic abnormality. There are 12 chapters describing different types of chromosome anomaly that can be carried by one of the parents, where there is a possibility of transmission of an unbalanced form to their offspring. Four further chapters discuss the counselling problems associated with chromosome abnormality in a child when both parents have normal karyo-