myotube 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

Prenatal Diagnosis in Obstetric Practice. Ed M J Whittle, J M Connor. (Pp 274.) Oxford: Blackwell Scientific Publications. 1989.

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

Chromosome Abnormalities and Genetic Counselling. R J M Gardner, G R Sutherland. (Pp 255; £26.00.) New York: Oxford University Press. 1989.

The primary aim of this book is to help cytogeneticists 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 cytogeneticists 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-

types. The first section of each chapter describes the biological facts underlying the type of chromosome abnormality, including descriptions of modes of segregation for structural abnormalities and explanation of the most probable outcome. The second section is dedicated to translating this information into advice for genetic counselling purposes.

A further three chapters concentrate on the counselling problems encountered during the course of prenatal diagnosis. The uninitiated are informed that "CVS prenatal diagnosis results are not as precise as those from amniocentesis". In this and many other aspects of interpretation of cytogenetic findings, this book can never be a substitute for past experience and a sound and up to date knowledge of published reports in a field where extra data are constantly being accumulated, but it provides an excellent starting point.

It will doubtless become a major genetic reference text for clinicians in all disciplines who are involved in counselling patients who have unusual karyotypes and will prove invaluable for cytogeneticists, whether experienced or training for examinations, in understanding and interpreting chromosome abnormalities.

M FITCHETT

X-Linked Mental Retardation 3. Ed J M Opitz *et al.* (Pp 707.) New York: Alan R Liss. 1988.

This volume contains the papers published in the May/June 1988 issues of the *American Journal of Medical Genetics*, published separately as the proceedings of the Third International Workshop on Fragile X and X-linked Mental Retardation (Troina, Italy, 1987). It will appeal to those interested in fragile X and other X linked mental retardation (XLMR) syndromes. However, only few persons or institutions are likely to wish to purchase this volume: those with ready access to the *American Journal of Medical Genetics* will not need to do so, while those who seek a comprehensive and contemporary review of the field may well be disappointed.

The conference report and the bibliography of XLMR are both of great

value. The clinical section comprises 28 contributions. There are seven interesting papers on non-fragile X XLMR, with particular emphasis on FG syndrome and the Golabi-Rosen syndrome. The other papers relate to fragile X and vary in their quality. They do contain important discussions of the manifestations of the condition in heterozygous females, and of the facial features of affected males, but many of the papers were of marginal interest to me. In no sense is this a review of the clinical picture of fragile X XLMR.

The next section covers prenatal diagnosis and treatment of fragile X. The four papers on prenatal diagnosis provide an honest account of the difficulties of prenatal diagnosis in this condition, as of 1987. Three papers record the effects of treatment of affected subjects with antifolic drugs (no help in cytogenetic diagnosis), with methylphenidate (possibly improves attention span in some), and folic acid (no apparent clinical benefit).

There are nine papers in the cytogenetic section, principally concerned with cytogenetic techniques and with correlations between cytogenetic results and clinical findings. The field is not reviewed, although some of the contributions are interesting. The molecular papers are inevitably out of date. Six papers are concerned with non-fragile X XLMR, and eight with fragile X. They are neither of great current interest to those involved in the field, nor do they provide an adequate review of the field to those not actively engaged in the molecular analysis. Six papers report population based findings. These are interesting but not new. There are four papers that speculate about the aetiology of the fragile X syndrome. These are also interesting, especially the arguments of Charles Laird.

As is inevitable in a book with this origin and format, the qualities of content and style vary enormously. Mental handicap libraries that do not subscribe to the book's parent journal, and persons with a major interest in XLMR but without access to the *American Journal of Medical Genetics*, may well choose to purchase this volume. Those who want a review of either the clinical or the molecular aspects of XLMR will need to look elsewhere.

A CLARKE

NOTICES

Medical Genetics: 1990

The Foundation for Advanced Education in the Sciences, Inc will present a three day course 'Medical Genetics: 1990' on 17 to 19 May 1990 in Bethesda, Maryland. The course will include didactic and problem oriented sessions. Topics will include gene structure and expression, chromosome structure, cytogenetics, reverse genetics, gene mapping, recombinant DNA techniques in medical genetics, endocrine genetics, neurogenetics, and the genetics of connective tissue disorders. Also included will be prenatal diagnosis, genetic counselling, and new approaches to the treatment of genetic diseases. The course is intended, in part, as a review for candidates for the examinations of the American Board of Medical Genetics, but will not ignore the excitement of current research. The course is approved for AMA category 1 credit. Further information is available from: Medical Genetics, FAES, One Cloister Court, Suite 230 Bethesda, MD 20814-1460, USA. Tel: (301) 496-7976.

Tuberous Sclerosis and Allied Disorders

This conference, sponsored by The New York Academy of Sciences and The National Tuberous Sclerosis Association, will take place on 23 to 25 April 1990 at the Hyatt Regency, Bethesda, Maryland. For further information contact: Conference Department, The New York Academy of Sciences, 2 East 63rd Street, New York, NY 10021, USA. Tel: 212-838-0230.

Genetics of Gastrointestinal Disorders

The 1990 March of Dimes Birth Defects Foundation Clinical Genetics Conference on 'Genetics of Gastrointestinal Disorders' will take place on 8 to 11 July 1990 at the Hyatt Regency, Dearborn, Michigan. For further information contact: Carol Blagowidow, Professional Services Department, March of Dimes Birth Defects Foundation, PO Box 2000, White Plains, NY 10602-9989, USA.