Book reviews

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La Pratique de l’Analyse Chromosomique

This small booklet, written in French, is the twelfth publication in the series "Techniques de Laboratoire". It consists of detailed descriptions of techniques and procedures routinely applied in cytogenetics laboratories. It is a good bench manual, especially useful to beginners in the field of human chromosome studies.

M Seabright

The Genetics of Neurological Disorders
Oxford Monographs on Medical Genetics No 9.
By Michael Baraitser. (Pp xii + 516; figures + tables. £35.00.) Oxford: Oxford University Press. 1982.

This book is a member of the excellent series of Oxford monographs on medical genetics and succeeds one of the same title by Dr R T C Pratt, who, in 1967, was one of the first to put some order into neurological genetics and whose book was both comprehensive and interesting. Since the publication of that book there have been great advances in the delineation of neurological genetic entities. At the same time, neurology has changed from a largely clinical discipline to one where scientific techniques predominate. For example, the use of histochemical stains and electron microscopy has revolutionised the classification of some muscular dystrophies, even causing some temporary confusion, since disorders described at the beginning of the century are reclassified if there are living descendants available for muscle biopsy! Also, the use of CT scanning has enlarged our knowledge of, and ability to recognise, disorders associated with cerebral malformations.

Dr Baraitser is a good choice to undertake the onerous burden of updating The Genetics of Neurological Disorders. He runs a genetic counselling service at the National Hospital for Nervous Diseases and is therefore familiar with the questions patients and their families ask, and he is Consultant Clinical Geneticist at The Hospital for Sick Children, and is therefore experienced in the recognition of syndromes.

Dr Baraitser has kept largely to the order and content of Pratt's earlier book but there are additional chapters and some have been expanded. The subjects covered include abnormalities and syndromes of the cranial nerves; malformations of the skull, spine, and nervous system; dementia; epilepsy; tumours; phakomatoses; and static and degenerative disorders of the basal ganglia, cerebellum, cerebellar pathways, spinal cord, peripheral nerves, and muscle. A paediatric neurologist will also find useful the sections on dyslexia, cerebral palsy, and degenerative disorders of childhood. The genetics of mental subnormality and psychiatric disorders have been dealt with in an earlier volume in the series.

Dr Baraitser states in the preface that "The main purpose of this book is to help with the identification of these (neurological) syndromes and to assist the clinician in the derivation of risks of recurrence". He succeeds well in these aims, particularly regarding the identification of syndromes. He has reviewed more neurological disorders than I care to think of: about the sum of reported experience of neurologists for a century or so! Many of these disorders have occurred in only a single family; the listing of them provides a comprehensive source of reference for clinicians struggling to classify an unusual constellation of signs. Empirical risks for genetic counselling are usually given, but there remains some uncertainty on how to counsel an isolated case. For example, how often is scapuloperoneal muscular dystrophy genetic and how often non-genetic? What is the chance that a male infant, severely ill with centronuclear myopathy, has an X linked variety? Although not stated in the preface, I consider this book to be directed largely to neurologists, for the clinical criteria by which a particular diagnosis is reached are not always given and many of the conditions are so esoteric that they are unlikely to come first to a clinical geneticist.

I was disappointed not to find Dr Baraitser's opinions on some controversial issues in this book. For example, I should like to have known his opinions on the following. What is the relative frequency of mutation in Duchenne muscular dystrophy in the two sexes? What proportion of patients with Huntington's chorea are new mutants? What is the explanation of the frequent paternal descent in juvenile Huntington's chorea, and the maternal descent in neonatal myotonic dystrophy? Is the predisposition to Parkinson's disease the result of monogenic, polygenic, or oligogenic inheritance? These topics are either not discussed or else the various opinions from different workers are listed. I would have welcomed Dr Baraitser's own interpretation of some of these issues.
I consider this an excellent successor to Dr Pratt’s book. Inevitably it is larger in order to maintain the high standard of being a comprehensive source of reference. Because it presents a clear and logical account of a complex subject it is tremendously helpful. It is essentially a book for neurologists and for them I strongly recommend it.

SARAH BUNDEY

Orthopaedic Problems in Inherited Skeletal Disorders
By Frank Horan and Peter Beighton. (Pp 142; 98 figures. £20-00, DM 85.) Berlin: Springer-Verlag. 1982.

This little book is well-produced with excellent illustrations. It is easy to read and (important) light to carry around.

The authors deal first in a very simple way with genetic principles; the second chapter describes in a general way the investigation and management of bone dysplasias, with one page allotted to antenatal diagnosis. Chromosomal disorders and polygenic inheritance with the common multifactorial disorders are not dealt with. Chapter three contains a full list of the international nomenclature, devised by the European Society of Pediatric Radiologists in Paris in 1969 and subsequently brought up to date. However, the classification adopted by the authors in discussing separate topics adopts the more usual and useful radiological divisions of predominantly disordered epiphyses, metaphyses, with or without vertebral involvement, decreased or increased bone density, etc.

This little book is best considered as a mini atlas and an introduction to the subject of skeletal dysplasias, rather than a technical guide to the postgraduate orthopaedic, plastic, or neuro-surgeon.

It is perhaps a little unrealistic to look for a volume containing the indications for and timing of surgical intervention with pre- and postoperative management in these rare and difficult disorders. It is only relatively recently that a few orthopaedic surgeons have turned their attention to this subject and there is as yet little material on which to base definitive decisions.

RUTH WYNNE-DAVIES

Clinical Genetics: Problems in Diagnosis and Counseling

This useful book reports the proceedings of the Twelfth Annual Birth Defects Symposium held in Albany in the autumn of 1981. The distinguished authors, all experts in their field, provide readable accounts of the current state of their art and this volume should prove valuable to anyone attempting to keep abreast of contemporary developments in the rapidly expanding world of medical genetics.

The book opens with an authoritative discussion of the vitamin supplementation studies on the prevention of neural tube defects, followed by a clear and concise overview of the ‘new genetics’ and its application in the antenatal diagnosis of the haemoglobinopathies. There are three papers with a biochemical flavour dealing with the difficult issues of duration of therapy in phenylketonuria (unresolved) and its management in pregnancy (which should commence before conception), plus the technical aspects of lysosomal enzyme assay and complementation studies with special reference to propionicaadiaemia.

There then follow four papers of cytogenetic interest. The first indicates that the risk of ‘severe anomaly’ in a fetus with an apparently balanced de novo structural rearrangement is unlikely to be great (three out of 41) and may not differ significantly from that for the general population. The need for more data is freely admitted. There is a helpful review of mosaicism and pseudomosaicism in antenatal cytogenetic diagnosis although the problem of trisomy 20 mosaicism remains. A study of ‘who gets amniocentesis’ is well worth reading, as is an excellent discussion of recurrence risks in Down’s syndrome, which offers useful guidance on how to cater for parental age, previous non-disjunction, and parental remarriage.

Chapters of general interest include a comprehensive account of the overgrowth syndromes, a review of empirical risk data in psychiatric disorders, and a brief but fascinating glimpse into the mind of Francis Galton, whose interests ranged from spectacles for divers to the statistical inefficacy of prayer (those who were prayed for most, such as royalty, tended to die youngest). It is a mixed comfort to note that Dr Opitz estimates that he can identify a cause and recurrence risk in only 20% of cases with an unknown syndrome. What hope is there for the rest of us lesser mortals? More depressing are the papers on cystic fibrosis, which reveal that despite considerable progress, reliable antenatal diagnosis is still some way off.

In presenting the proceedings of any symposium the editors and publishers face a difficult task in maintaining literary and technical quality without sacrificing speed of production. In this volume they have largely succeeded. The text is of a high standard,