Book reviews

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Neurofibromatosis: Phenotype, Natural History and Pathogenesis

By Vincent M Riccardi and June E Eichner. (Pp 305; figures+tables. £39.50.) Baltimore: Johns Hopkins University Press. 1986.

The clinical material for this monograph is drawn from the extensive clinical experience of Riccardi in the assessment and follow up of neurofibromatosis (NF) patients by the NF programme at the Baylor College of Medicine, Houston, which was established in 1978. This material is combined with excellent clinical photographs and an exhaustive review of published reports to give a detailed overview of all aspects of the disease.

Readers should not be deterred by the first chapter of the book which describes the methods of evaluation used by the Baylor NF programme. This was found to be rather heavy going and much of the detail may have been better as an appendix. The remainder of the book, however, is well written and every aspect of the disease is considered in detail. The chapters on other forms of NF and the pathogenesis of NF are particularly good. The appendix summarises the findings of the evaluation of 238 patients and provides useful data for those interested in designing a follow up programme for NF patients; many of the investigations originally undertaken by Riccardi have been shown to be unnecessary.

The book will be a valuable source of reference not only for medical geneticists but also for all the other medical specialists involved in the treatment of NF and its complications. It also provides a useful starting point to scientific and clinical investigators undertaking NF research: the areas that the authors feel deserve further investigation are highlighted at the end of each section.

SUSAN M HUSON

Self-Assessment in Medical Genetics

By J M Connor and J R W Yates. (Figures+tables. £6.80.) Oxford: Blackwell Scientific Publications. 1986.

Each right hand page of this little paperback shows two photographs or case histories and poses a series of questions: What is the likely diagnosis? What is the prognosis? What is the recurrence risk? etc. To

find the answers, turn the page. Answers include brief explanations, but no references. A 'Further Reading' section at the end gives some book titles, modestly relegating the Glasgow team's excellent 'Essential Medical Genetics' to the Genetic Counselling section.

Inevitably the pictorial format leads to some concentration on malformations to the detriment of purely biochemical or functional disorders. Nevertheless, there are questions on non-specific mental retardation, grand mal epilepsy, congenital deafness, and similar important practical problems. Pedigrees, karyotypes, and DNA polymorphisms also provide visual interest, so that overall the book reasonably well fulfils its claim to show conditions illustrating recent advances, conditions with treatable complications, and conditions likely to be encountered in everyday practice.

If the price were three times as high, I would complain at the quality of the photographs, which are adequate rather than beautiful. In one or two I found it hard to make out the feature of interest, but at the price they are satisfactory. I did not spot any blunders; the crisp format makes for rather didactic statements, not out of place in an examination oriented book.

A subject which is very unevenly taught in medical schools, and which is advancing with unprecedented speed, must be a good one for a self-assessment text. This is a modest book with no pretensions to give the reader a deep grasp of genetic principles, but it is cheap and well laid out. The expected market is among candidates for MRCP or MRCOG exams, though the opportunity to try a little quiet self-assessment may well appeal to the many self-taught people already established in clinical genetics. But buy it quickly before too many of the answers suffer the fate of No 28c: Is prenatal diagnosis of cystic fibrosis possible?

ANDREW P REA

Ethnic Differences in Reactions to Drugs and Xenobiotics

Progress in Clinical and Biological Research Vol 214. Edited by W Kalow, H W Goedde, and D Agarwal. (Pp xv+583; figures+tables. £64.00.) New York: Alan R Liss. 1986.

The work of Dr Kalow in pharmacogenetics has

been well known for many years, and in this volume he and his colleagues turn their attention to an important section of the field that has been less studied than it deserves—population differences in inherited drug response. Yet a few obvious pointers have long indicated its intrinsic interest—the population differences in frequency of the drug induced haemolysis of G6PD deficiencies, the low frequency of slow acetylators of isoniazid among the Eskimo, the response to alcohol in Orientals—and today it is clear that identical reactions to a given drug in all populations at identical frequencies are most unlikely. The lack of attention is perhaps to be attributed partly to the complexity of assay and the difficulties of obtaining and persuading families, particularly those of non-European culture, to participate in procedures of loading and sampling that may require several days.

In this volume are collected 36 of the papers presented and summaries of the discussions at a conference on this topic held in October 1985. In this first section, some fundamentals are established. Goedde gives a useful summary table of the different drugs to which variant responses are known, the population incidences, and the mode of inheritance. Nei compares the relationships of populations as indicated by the blood polymorphisms with the distribution of lactose tolerance, aldehyde dehydrogenase-1 deficiency, and pseudocholinesterase variants, to suggest that the first two of these polymorphisms have existed for a very long time, but that the third is maintained by balance between weak selection and mutation. Anderson *et al* point out that diet needs consideration, for it may influence the metabolism of drug substrates, and they show that dietary change can substantially alter the metabolism of steroid hormones.

Section 2 concerns the drug metabolising enzymes and includes family studies (for example, of the serum paraoxonase/arylesterase polymorphism), ethnic comparison (for example, of ethanol oxidation), and evidence of further heterogeneity (for example, a different debrisoquine metabolising mutant in Ghanaians from that in Europeans and the indication that apparent homozygous poor metabolisers in Ghana are heterozygotes for the D^L and D^{Gh} alleles). There is an excellent review of the acetylation polymorphism by David Price-Evans.

The third section concerns the actions and fates of drugs and chemicals, such as primaquine, phenytoin, and ethanylestradiol. Again, ethnic differences appear, for example, in the response to certain hypertensive agents between blacks and whites, while the differences in phenytoin elimination rates suggest that the Eskimo run the risk of insufficient anti-epileptic control. Section 4 on protein variants

with potential pharmacogenetic consequences deals both with plasma binding proteins (for example, albumin and orosomucoid) and with receptor proteins to growth hormone, androgen, and insulin, for example.

Some methodological problems in studies of population variation are reviewed in section 5. The final papers on implications for therapy by Price-Evans and for occupational and environmental exposure to chemicals by Omenn provide a fair appraisal of the potential benefits to be derived from pharmacogenetic studies.

This is an interesting book, containing some first class papers which are well worth reading, although there are also a few which are not. It is unfortunate that the standard of production, with its varying type faces, varying format, varying type intensity, poor proof reading, illegibility of legends to some figures, and poorly laid out tables, is so appalling.

D F ROBERTS

Cholinesterase

By M Whittaker. (Pp 97; figures+tables. £40.70.) Exeter: Karger. 1986.

This slim volume, 97 pages of text, is number 11 in the Monographs in Human Genetics series edited by Professor Lars Beckman. It forms a welcome addition to the series, which last had issue about eight years ago, and represents the distillation of Dr Mary Whittaker's expert knowledge on the subject of human cholinesterase. The text is well organised with 10 short chapters, each subdivided and fully illustrated with charts and diagrams and tabulated information. More than 500 original references are cited and their alphabetical arrangement and the provision of full titles and authorship is most useful. There is also an excellent comprehensive index with more than 300 entries.

This book contains everything anyone would wish to know about 'pseudo' cholinesterase (acylcholine acylhydrolase, EC.3.1.1.8) not to be confused with the 'true' or 'specific' acetylcholinesterase (acetylcholine acetylhydrolase, EC.3.1.7.7). The distinction between these two enzymes is made in the very first section of the book and the text thereafter is concerned exclusively with cholinesterase (EC.3.1.1.8). This unfortunately means that there is no mention of cholinesterase in amniotic fluid and the determination of acetylcholinesterase (EC.3.1.1.7) as a means of prenatal detection of open neural tube defects. This book will therefore be most useful to chemical pathologists, anaesthetists, and clinical geneticists faced with the occasional case