Neurofibromatosis: Phenotype, Natural History and Pathogenesis

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

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 is adequate rather than beautiful. In one or two cases, I found it hard to make out the feature of interest. Even at the price they are satisfactory. I did not spot any blunders; the crisp format makes for rather schematic 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 quiet little 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 Read

Ethnic Differences in Reactions to Drugs and Xenobiotics

The work of Dr Kalow in pharmacogenetics has