A Clinician's View of Neuromuscular Diseases

This book, which has now reached its second edition, gives clear and detailed clinical descriptions of neuromuscular diseases. Any book whose introduction starts with an apology that it is not an 'Encyclopaedic Compendium' but rather a practical guide for the practising clinician is to be welcomed. The style is very readable and I am sure that it will be, and indeed has been, used by neurologists, paediatricians, and clinical geneticists on a day to day basis.

The chapter on symptoms and signs gives an accurate and concise guide to the assessment of the patient with neuromuscular disease. The description of the deterioration of the boy with Duchenne dystrophy is very valuable as is the author’s precis of all the research into the causation of this sad disease. In this chapter the genetic aspects are well covered with a realisation that research is likely to overtake present advice on carrier detection and prenatal detection. Nevertheless advice on genetic risks is rather harder to find in some other sections of the book.

This book is a good guide to the physical management of children with muscle disease; however, there is little on the home adaptations which make such a difference to the lives of patients and their families. There is also little on educational options for these children. The presentation of the book is spoilt slightly by the lack of paragraph headings which makes it difficult to find information quickly in some of the longer chapters. Despite these rather minor points this is a splendid book which will be of use to the practising clinician. There are few fields where accurate diagnosis is as important as neuromuscular disease in giving genetic advice. This book makes that accurate diagnosis easier and will therefore be of particular value to the clinical geneticist.

J R Sibert

The Man Behind the Syndrome

Physicians and surgeons have had bibliographical details of their notables enshrined in various texts for several years, and now geneticists have theirs. Here are presented details of those who have achieved ".... eponymous immortality or notoriety" for disorders and syndromes which have a significant genetic or chromosomal basis. The book is divided into two sections. In the first, a photograph or portrait is provided for each of 100 persons, most of whom are dead, or in the case of Klein, Rieger, and Wiedemann have "...reached such seniority that professional jealousy is unlikely to be aroused in their colleagues". Few would take issue with the inclusion of any in this section, of whom incidentally only three are women (Julia Bell, Cornelia de Lange, and Gertrud Hurler). The mean age at death in this group is 71 (SD 12) which suggests that having one’s name eponymously associated leads to good survival! There are, however, notable exceptions: Laurence (of Laurence-Moon syndrome) died at 42 and Pompe was executed at the age of 44 for being a member of the Dutch resistance movement.

The second section contains brief bibliographical information on 110 others, many of whom are still active and some might doubt if all these suggested eponyms will survive the passage of time.

Professor Beighton and his wife have produced scholarly work which is delightfully written and well referenced and will be especially valuable to those with interests in the history of genetics.

Alan E H Emery

Malformations in Children from One to Seven Years:
A Report from the Collaborative Perinatal Project

This book is a report on a longitudinal study of congenital malformations in children up to the seventh year of life. There was a previous report after one year on the same cohort. Ascertainment of the children was from 12 institutions throughout the USA, in the Collaborative Perinatal Project of the National Institute of Neurological and Communicative Disorders and Stroke. The sample size was 5229 consecutive single births with known outcomes including fetal and neonatal deaths. The first third of the book is devoted to criteria and definitions of sources of data, plan of the work, and discussions of malformations detected from one to seven years and comparison with those detected in the first year study. Racial and sex differences are examined. The section on malformations detected after the age of one year is of interest, as is the follow up information on children found to have a particular malformation at one year, since this gives some information on prognosis. Two-thirds of the book is devoted to detailed appendices where the number of individual malformations are given and the...