
Professor Wiedemann and his associates have contributed greatly to the delineation of syndromes over several decades, and this atlas reflects their experience and the value of careful documentation and follow up. The atlas is now in its second English edition and each entry follows a format with text on one page facing illustrations opposite. The text is telegraphic in style divided for each syndrome into nosology, sign and symptomatology, investigations, aetiology, frequency, course, treatment, and references.

Many of the illustrations are old and some are taken against a dark background; consequently some appear indistinct and it is difficult to see abnormalities of skin texture or pigmentation. Some well delineated syndromes are not illustrated, such as VATER, TAR, L, E, Pena-Shokeir, and CHARGE association, though I did wonder whether the "unknown" syndrome on page 44 could be this latter disorder. The inclusion of the authors' 'unknowns' spread throughout the atlas is unsystematic and does not help the organisation of the book, which is not explicit anyway.

This atlas, although enlarged in this edition, has not been extensively rewritten and as a consequence recent knowledge about the genetic basis and mechanisms underlying some of the syndromes is not included. For example, most imperfections types IIA-C are referred to as usually recessive disorders whereas recurrences in some families have now been shown to be the result of parental germinal mosaicism.

Most dysmorphologists like to have all the available 'syndrome books' to see different cases illustrated at varying ages. Although this atlas would not be my first choice, it is a good value, reasonably comprehensive, and constitutes a helpful contribution to the body of publications.

DIAN DONNAI


The recent advances in understanding some of the ataxias exemplify the different ways in which genetic and other studies can help to elucidate the mechanisms of disease. This interesting volume presents many new and exciting aspects of ataxic conditions, some of which are not yet fully explained. It is an enjoyable and stimulating book.

First there is a clear account of the classification of ataxic disorders, which emphasises the recognition of those few syndromes that are treatable. An improved delineation of cerebellar syndromes can be made with modern imaging, for MRI techniques show how the cervical cord is atrophied in patients with Friedreich's ataxia while the cerebellum appears normal, whereas in early onset spas tic ataxia, the cervical cord is normal but there is atrophy of all parts of the cerebellum. In adult onset ataxia type 1 (ADCA type 1) MRI scanning shows atrophy of the entire cerebellum together with atrophy of thepons, middle cerebellar peduncle, medulla, and upper cervical cord. However, MR findings in pure cerebellar atrophy (ADCA type III) show atrophy confined to the cerebellum. Patients with the spastic ataxia associated with neuropathy (Charlevoix-Saguenay atrophy) of the superior cerebellar vermis and atrophy of the cervical and thoracic segments of the spinal cord.

Gene mapping has been successfully accomplished in Friedreich's ataxia, ataxia telangiectasia, and some families with ADCA type I. In Friedreich's ataxia there is substantial evidence that only one gene locus is involved; its localisation is a tribute to international cooperation and a lot of hard work and it was the third disease gene locus to be identified. However, it is proving difficult to pinpoint the gene more accurately, owing to its position near the centromere on chromosome 9 and the un informativeness of markers used in the early studies. Recent identification of CpG doublets (which usually lie alongside coding sequences) and the use of yeast artificial chromosomes to clone segments of DNA from the region are valuable strategies. Moreover, Chamberlain et al are screening cDNA libraries obtained from fetal and adult brain and cerebellum. The prospects for finding and sequencing the gene look promising. Ataxia telangiectasia is an interesting condition in which the type of gene that could cause such varied effects on the immune system and on chromosome stability, with ataxia and ataxic neuropathy being the two commonest features. In humans the disease may be inherited as an autosomal dominant (five point mutations and two insertions in the prion protein gene have been described) or as a rare infection with an aberrant prion protein which then catalyses a post-transcriptional event so that further abnormal prion protein is derived from the host's genetic sequence or, most commonly, by an unknown mechanism different from the first two. It is interesting that the host's genotype plays some part in the manifestation of disease. In inherited prion protein disease, homozygosity for a polymorphism of codon 129 leads to early onset of disease, while the few patients who have developed Creutzfeldt-Jacob disease after being given cadaveric derived growth hormone are predominantly homozygous for valine at this position. Patients with sporadic Creutzfeldt-Jacob disease are more often homozygous for either valine or methionine at codon 129 of the prion protein and would be expected from the distribution of these polymorphisms in the general population.

It used to be thought that the neurodegenerative disorders formed a depressing group of conditions because no prevention or treatment could be envisaged. However, this book illustrates that such a view is no longer true and that there are great possibilities for the future at least in regard to the ataxic conditions. This volume, therefore, in the series of Advances in Neurology, is of great interest to neurologists and to those geneticists interested in neurologic diseases.

SARAH BUNDEY


A useful addition to this series, Genetic Engineering provides an excellent introduction to the technological wizardry behind the science of molecular genetics. Little previous knowledge is assumed and the text