Book reviews

Journal of Medical Genetics 1987, **24**, 317–320

Genetic Counseling for Clinicians

By Kenneth L Garver and Sandra G Marchese. (Pp 356; figures + tables. £38.00.) Chicago: Year Book Medical Publishers. 1986.

This book begins with the laudable aim of providing "a comprehensive and easily understood book on genetic counseling for primary care physicians, nurse practitioners and social workers". I am sorry to say that, in my opinion, it fails to achieve this aim and cannot be recommended.

The first five chapters cover basic genetics, reproductive options, and prenatal diagnosis. There follow 14 chapters on specific disorders, mostly organised along 'system' lines. The style is didactic and rather forbiddingly dogmatic. The chapter on inheritance patterns, for example, begins with a glossary and continues with a series of baldly stated rules. Many of the 'system' chapters are entitled 'Patients at risk for . . . disease', but the overwhelming impression is of a catalogue of diseases with little or nothing about the clinical evaluation of patients and their families and the ways in which they cope with inherited disorders. This impression is strengthened by the presentation of much material in tables, some covering several pages. Common and rare disorders are given comparable amounts of space with no indication of relative frequency. The one thing that would make a listing of rare disorders useful, a reference for each one, is not provided.

There are some alarming omissions. The table on 'dwarfism' lists 13 conditions but does not mention osteogenesis imperfecta, hypophosphatasia, spondyloepiphyseal dysplasia, and pseudoachondroplasia. I was unable to find any mention of the fragile X syndrome or X linked mental handicap in the book, including the index, nor is there any coverage of the diagnostic evaluation of a patient with mental handicap. DNA analysis is mentioned fleetingly in connection with sickle cell anaemia, the thalassaemias, and Huntington's chorea, but the methods and underlying principles are not explained. One would not know that only a linked marker, with all its attendant limitations, is available for Huntington's chorea, and DNA diagnosis in X linked disorders is not mentioned at all. In the sections on chromosomes there is no Paris Conference diagram and no explanation of banding notation.

Reading this book was a disappointing experience. Other authors have shown that a book of this size can provide a guide to the field of clinical

genetics in appropriate depth for newcomers, and at the same time convey a sense of intellectual excitement and unanswered questions. I hope that the distinguished authors and publishers, together with the 21 eminent persons listed as having had a hand in it also, will radically re-think their approach for the second edition.

N R DENNIS

Ataxia Telangiectasia: Genetics, Neuropathology, and Immunology of a Degenerative Disease of Childhood

Edited by R A Gatti and M Swift. (Figures + tables, £60.00.) New York: Alan R Liss. 1985.

Ataxia telangiectasia (A-T) is a fascinating disorder and has been the subject of a growing amount of investigative work over the past 10 years. The patients are characterised clinically by a progressive cerebellar ataxia presenting in early childhood. Consistent laboratory markers in these patients include a raised level of serum alpha-fetoprotein, a defect in cell mediated and humoral immunity, an increased level of spontaneously occurring chromosome rearrangements in peripheral lymphocytes, and an increased radiosensitivity in cultured cells. There are reports suggesting that different types of ataxia telangiectasia may be recognisable and genetic complementation has been demonstrated at the cellular level.

The importance of A-T for the laboratory scientist is as a model system for investigating cellular changes accompanying the development of malignant disease. A-T patients have a greatly increased likelihood of developing lymphoid malignancies in particular.

There are of course many unanswered questions regarding A-T. What is the nature of the suggested DNA repair defect in these patients' cells? What is the relationship between the specific chromosomal translocations in A-T lymphocytes and T cell leukaemia? Where is the A-T gene located in the genome? Is there a means of detecting carriers of the A-T gene and do these people also have an increased risk of cancer?

The editors of this book have brought together contributions made at a conference with contributions to update or expand the content. The first chapter by Dr Elena Boder gives a good introduc-