Ataxia-Telangiectasia. A Cellular and Molecular Link Between Cancer, Neuropathology, and Immune Deficiency

Edited by B A Bridges and D G Harnden. (Pp xix + 400; figures + tables. £20.75.) Chichester, New York: Wiley. 1982.

In essence this book contains papers presented at a workshop held in November 1980, a workshop that appears by these accounts to have been a very exciting meeting. However, the book is more than this, for the editors have asked each author to give a brief review of the work leading up to their research, before presenting their results, and in addition have included six excellent review chapters. The result is that the book has become a monograph and a very stimulating one, concerning one of the most interesting diseases that exists.

Ataxia-telangiectasia (AT) is an important disease to study because as well as being characterised by progressive neurological degeneration and abnormalities in blood vessels and in the immune system (Waldmann, p37), it confers a susceptibility to certain cancers both in the homozygous recessive state (Spector *et al*, p103) and in the heterozygous state (Swift, p355). This susceptibility could well be related to the increased sensitivity of chromosomes to X or Y irradiation (Taylor, p53), to the formation of chromosome clones, and/or to the increased sensitivity of cultured fibroblasts and lymphocytes to X or Y irradiation (Lehmann, p83). This latter response has been carefully analysed by Cox (p141), whose experiments show that in AT there is a complete block in the repair of potentially lethal radiation-produced damage at both high and low doses of radiation, and that different AT biochemical phenotypes appear to be equally deficient. The presence of this DNA repair defect would account for the devastating response that AT patients have when given therapeutic radiation for cancer, and further study of their defects will be useful in the future understanding of normal persons' responses to radiation. The abnormal response of blood and skin cells to irradiation also enables antenatal diagnosis of the condition in at least some cases and this is discussed by Gianelli *et al* (p393).

The sites at which chromosome breaks and rearrangements occur are not random and those particularly involved are 7p14, 7q35, 14q12, and 14q32 (Hecht and Kaiser-McCaw, p235; O'Connor *et al*, p259). This raises the possibility that one of

these sites either represents the position of the recessive AT gene or of other genes concerned with carcinogenesis or immune deficiency.

There is a further puzzle about AT which emerges from this book. Paterson *et al* (p271) and Inoue *et al* (p305) describe how cultured hybridised cells from different patients may 'complement' or 'make good' the DNA repair defects that are present in either when cultured alone. Several different complementation groups, based on such hybridisation experiments, exist for AT which suggest that there are different genes for AT, situated at different loci; indeed it is difficult to think of an alternative explanation for these laboratory findings. However, this hypothesis is not compatible with the population findings. As Hecht and Kaiser-McCaw (p197) observe, the consanguinity rate in AT is low, too low indeed for a rare condition that really consists of several even rarer entities. The commonly quoted estimate of the frequency of AT is based on its prevalence in Los Angeles school children and gives a figure of about 1 in 40 000. The observed consanguinity rate of about 3% is just compatible with this, but not with the suggestion that AT consists of several rarer entities caused by non-allelic genes. It would be useful if a further prevalence study could be carried out.

In summary, this book gives a comprehensive account of a disease that is important because of what it can teach about carcinogenesis, neurological degeneration, and immune deficiency. It should serve as a basis for future research ideas and actions. I can recommend it not only to those clinicians and scientists who know one aspect of AT, or a little of all aspects, but also to those who are unfamiliar with the subject.

S BUNDEY

Heritable Disorders of Connective Tissue

American Academy of Orthopedic Surgeons Symposium, San Diego, California, 1980. Edited by W H Akeson, P Bornstein, and J Glimcher. (Pp xiv + 380; 193 figures + tables. £43.50.) St Louis, Toronto, London: Mosby. 1982.

These are the beautifully produced proceedings of a symposium held in 1980 and published in 1982. It contains a very adequate summary of the molecular biology and protein chemistry of the inherited defects of collagen, but also includes much useful

information on other connective tissue proteins such as the proteoglycans and phosphoproteins. Although the application of recombinant DNA techniques to inherited collagen defects is now accelerating, the chapter by Tolstoshev and Crystal is remarkably up to date and provides an accurate guide to anyone interested in this field. The contributors include most of the recognised American investigators in the collagen field with a few notable exceptions.

This work will be of interest to relevant workers in the field including molecular biologists, medical geneticists, paediatricians, orthopaedic surgeons, rheumatologists, and metabolic physicians. Unfortunately at £43.50 most people will read a library copy rather than purchase their own, although perhaps a cheaper soft back edition would have overcome this problem.

F M POPE

Issues and Reviews in Teratology

Volume 1. Edited by Harold Kalter. (Pp xvii + 354; figures + tables. \$45.00.) New York, London: Plenum Press. 1983.

Editors and publishers must think carefully before launching a new series of review volumes on an oversaturated readership. Volume 1 of *Issues and Reviews in Teratology* leaves one less than convinced of a need for such an enterprise.

The contents of the book reflect its title and, rather like the Curate's egg, I can assure you that parts of it are excellent. Chapter 4 by Saxen, a cameo in epidemiology, is well worth reading. Entitled 'Twenty Years of Study of the Etiology of Congenital Malformations in Finland', it summarises the strengths and weaknesses of a reasonably comprehensive regional attack on the problem. Hendriks in 'Developmental Toxicity and Non-human Primates: Interspecies Comparisons' introduces the general teratologist to the mystique of the non-human primate and compares its teratogenic response to a number (albeit a very small number) of other mammals. Schardein takes another look at 'Teratogenic Risk Assessment: Past, Present and Future' which differs from the endlessly repeated legal requirements in various countries in making suggestions for the future. One need not necessarily agree with his predictions.

The reader will be grateful to Dr Snow, 'Restorative Growth in Mammalian Embryos' and to Drs Grabowski and Daston, 'Functional Teratology of the Cardiovascular and other Organ Systems' for introducing the new ideas suggested by the titles of their stimulating papers. A good 'in depth' account

of our present knowledge of the mechanism of acetazolamide teratogenesis is given by Drs Hirsch and Scott and Dr Theisen attempts to knock another nail into the coffin of the neuropathic theory of limb reduction defects—a difficult task because it has been done so often before.

Readers of the *Journal of Medical Genetics* will find little new in Dr Carrs' 'Cytogenetics of Human Reproduction Wastage' or Dr Boué and colleagues' 'Genome and Chromosome Mutations: Balance between Appearance and Elimination'. There is some repeated common ground here but the non-geneticist might choose either of these from the many competent reviews currently available.

Drs Nishimura and Warkany, both doyens of teratology, each give us a regrettably brief glimpse of their accumulated wisdom.

F BECK

Blood Relations. Blood Groups and Anthropology

By A E Mourant. (Pp vi + 146; figures + tables. £15.00.) Oxford: Oxford University Press/Clarendon Press. 1983.

Dr Mourant's invaluable compilations *The Distribution of the Human Blood Groups* (1954, 1976), *The ABO Blood Groups* (1958), *Blood Groups and Diseases* (1978), and *The Genetics of the Jews* (1978) are well known for what they are, authoritative statements of data published and unpublished, screened for technical reliability, and all subjected to uniform methods of gene frequency calculation, on gene frequencies in human populations throughout the world. They are the classic sources to be consulted for any description of gene frequency distribution, be it in country or continent, or for analytical purposes, to provide normal series against which special samples can be compared. The present book takes a more leisurely look at the interpretation of the data in these volumes, and harks back to Dr Mourant's first essay in compilation in 1954, when he endeavoured to interpret in anthropological terms the patterns discernible in the much more restricted material then available.

Following a short introduction on how we recognise people, which points out the differences between recognising populations and recognising individuals, and the usefulness for the former of simply inherited serological characteristics, there is a chapter on elementary genetics. Partly historical, it also deals with gene frequencies and the processes that change them, the storage of genetic information, linkage, and the principal blood group, serum protein, and enzyme polymorphisms that show sufficient gene frequency variation for them to be included in