Book reviews

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Molecular Basis of Inherited Disease

By K E Davies and A P Read. (Pp 77; £5.95.) Oxford: IRL Press. 1988.

I enjoyed reading this book. It is clearly written in a crisp, concise style that occasionally verges on the terse. The aim is to review recent advances in our understanding of the molecular basis of human inherited disease. There is a glossary of genetic diseases and another of scientific terms, but some acquaintance with the terminologies of Mendelian genetics, of molecular biology, and of biochemistry would be advisable.

The first chapter briefly reviews pedigree structures and the nature of genetic markers. The second chapter outlines the techniques used to map disease loci and random DNA markers, and to identify gene mutations at the RNA level and by using the polymerase chain reaction. This chapter includes a discussion of genetic linkage analysis. The third chapter examines methods for the identification and isolation of disease genes. The techniques of subtractive hybridisation, of genomic enrichment by chromosome sorting, the pERT methodology, and chromosome mediated gene transfer are described. Large fragment gel electrophoresis is discussed and its uses in the construction of jumping libraries and of physical maps and thence in gene identification.

The fourth and final chapter focuses on the nature of the mutations found in different human genetic diseases. Attention is paid especially to β thalassaemia and to Duchenne muscular dystrophy, but reference is also made to a variety of other conditions.

This book sketches the approaches being adopted in the study of human genetic disease and the type of answers being found. It spells out some of the applications of these methods that have been fruitful, and indicates in passing where there is potential for future work. I would recommend this book warmly to students or researchers with some knowledge of molecular genetics but little knowledge of human genetic disease, or vice versa. It will be of use to all those who want a concise summary of the field, and should enable the reader to follow current publications. The text is almost free of errors of typography, and the clarity of the layout and illustrations is only limited by the high density of facts per cm². The book does not set out to be a

practical guide, nor does it attempt to provide sufficient detail to satisfy those who need to understand in depth the molecular pathology of specific diseases.

ANGUS CLARKE

Human Mating Patterns

Society for the Study of Human Biology Symposium 28. Edited by C G N Mascie-Taylor and A J Boyce. (Pp 237; £25.00.) Cambridge: Cambridge University Press. 1988.

The 14 papers in this small book were presented at a two day meeting of the Society for the Study of Human Biology at Oxford in April 1986. The broad topics are the factors determining mating choice and the biological consequences of different mating patterns. Papers are grouped under the headings Historical and Demographic Studies, Mate Choice and Assortative Mating, Medical and Biological Aspects of Inbreeding, and Social, Religious, and Cultural Factors.

Clinical geneticists looking for an easy digest of the relevant (to them) bits of anthropology will not find it here. Most of the papers discuss detailed data or methodology, and often assume familiarity with the references. The most promising title, *Mating Patterns and Genetic Disease*, heads a chapter by Harper and Roberts which explains (very nicely of course) to anthropologists matters well known to clinical geneticists.

Inbreeding is the main topic which obliges clinical geneticists to think about mating patterns. Bittles and Makov give a useful summary of published reports on the incidence of stillbirths and deaths in the first year in inbred marriages. If it were true, as often claimed, that the average person carries two to four lethal genes, the costs should be much higher than they really are. Why then did the classic studies of Morton, Crow, and Muller give this high number of lethal equivalents? Bittles and Makov suggest reasons, but it would have been nice to see more discussion of this problem. Meanwhile the zoologists Read and Harvey (*Genetic Relatedness and the Evolution of Animal Mating Patterns*) suggest that animals might maximise their inclusive fitness by

mating with first or second cousins rather than outsiders. An interesting sidelight is given by Jakobi and Darlu whose paper on Mating Patterns in Isolates considers the POG. This is the Probability of Origin of Genes in latter generation members of the isolate. I had not appreciated how variable the contribution of the founders could be, and how small, given even a very few outside marriages.

Several papers look at the role of geographical, social, and cultural factors in mate choice. The authors occasionally go to some trouble to prove things I thought I knew already: that you are more likely to marry someone from down the road than someone from over the hill and far away, or that poor girls who wish to marry rich men should be beautiful. Perhaps those who understand more of the subject appreciate details which escaped me. Assortative mating is considered by several authors, with useful data on physical and psychological traits. A more difficult question is whether different degrees of assortative mating are associated with differential fertility. Here the evidence is inconclusive except for years of schooling, which has a low genetic content.

In summary, reading this book probably won't answer your questions about the different mating patterns now seen in important parts of the British population. It will show you what biological anthropologists do, and give numerous curious titbits of information. The paper I most enjoyed had nothing to do with genetics. This was an elegant exposition of the connection between romantic love, capitalism, and the demographic transition. If you won't let your family choose your spouse, then you can't expect them to support you when you marry. Therefore you must accumulate your own wealth and delay marriage until you can afford it. You can make more money when single, so those who value money above love never marry. All this means fewer children and less need for war, famine, or pestilence to keep the numbers down. I am not sure whether it is love or capitalism we should thank for this happy state, but it's not genetics.

ANDREW P READ

Ceroid-Lipofuscinoses. Batten Disease and Allied Disorders

Edited by J M Opitz, R K Pullarkat, J F Reynolds, and L M Spano. (Pp 308; \$85.00.) New York: Alan R Liss. 1988.

A literary weekly recently ran a competition in which readers had to suggest a book title of such stupefying obscurity that not a single copy would be

sold. As I recall, the winning title was *A history of the theory of rain*. The title of this book would surely have run a close second at least; it is quite easy to find distinguished professors of genetics who have never heard of the ceroid-lipofuscinoses let alone seen a case. The contents of this excellent volume are a marvellous illustration of how misleading the title and cover (dull green) of a book can be.

It is an account of the proceedings of the International Conference on Ceroid-Lipofuscinoses held on Staten Island, New York over two years ago (May 1987). As such it is a model of how such proceedings should be produced: impeccably illustrated, organised, and edited. The ceroid-lipofuscinoses are probably the most common of the neurodegenerative disorders of childhood. The common phenotype is exceptionally cruel in its relentless and devastating effect on child and family. By comparison cystic fibrosis seems almost the work of a benign providence. In spite of 20 years of intensive investigation its biochemical basis remains a tantalising enigma. Promising theories for its causation have perished with the same inevitability as its victims.

The volume opens with an invaluable and comprehensive bibliography for the years 1970 to 1986. The 27 papers presented at the conference have been cast as chapters arranged in four sections: Clinical and pathological studies, Animal models, Biochemical studies, and Treatment. The Riders, whose Children's Brain Disease Foundation has done so much to foster research in Batten disease, provide a useful overview of recent work and future hopes.

The individual contributions are of a uniformly high standard and range from careful accounts of long term clinical trials to some fairly abstruse biochemistry. Excellent accounts of the features of the various subtypes will fascinate the paediatric neurologist. A short but masterly account of the Newfoundland aggregate of NCL will intrigue the geneticist. Comprehensive descriptions of the several animal models available will satisfy the most demanding veterinary pathologist. An up to date review of current biochemical investigations including changes in lymphocyte membrane fluidity, cathepsin B activity measurements, and alterations in dolichol linked oligosaccharides will mystify all but the most erudite biochemists. The accounts of the various ingenious approaches to treatment are models of careful and cautious interpretation.

Inevitably the delay in publication engendered by production of such high quality conference proceedings dictates that the latest work and approaches to this disease are not discussed. Identification of the proteolipid subunit of ATP synthase as a