AN INTRODUCTION TO MEDICAL GENETICS. J. A. F. Roberts and M. E. Pembrey. Oxford University Press, 7th Edition, 1978. Pp. xviii+324. Price £5·50.

In 1940 Fraser Roberts wrote the first edition; it was designed for physicians, and deviated little from man, explaining what was known with little help from plants, flies or mice. Nineteen years later a second edition included blood groups and linkage, and a particularly lucid description of the rhesus system. Now, after a further 19 years, and 8 years after the sixth edition, this edition has acquired further chapters on haemoglobin, further additions on blood groups, tissue antigens, chromosomes and foetal diagnosis, and a further author. It retains most of its original words and diagrams and, with them, its clarity of thought and elegance of expression. The additions over the last 38 years seem surprisingly small, and the need for omission even less.

The outstanding omissions are any very adequate discussion or diagram on protein synthesis, and little on immunoglobulins or the enzyme variants, in spite of an excellent summary of the haemoglobins and HLA. The chromosomes are well covered, although the statement that foetal nuclear sexing is confirmatory, rather than definitive, is unfortunate. Intelligence is entirely omitted, as in previous editions. Multifactorial inheritance is used as a synonym for polygenic inheritance; most users of this word include environmental factors. Not only is the environment apparently regarded as a minor influence on this reputed class of disease, but the " polygenes " continue to be invoked as causal agents. The senior author's own work on blood pressure is hardly mentioned. This book retains its lead in clarity of expression and elegance of prose, and the publishers have retained their lead in excellence of reproduction of text and diagram, without any unreasonable penalty in purchase price.

Age shows its effect not in any inadequacy, but in a certain gauntness, and an overemphasis in importance, and in severity, of those conditions whose milder and commoner forms were not readily diagnosed in the past. The disorders presented are mostly atypical in their severity and, while suited to illustrate pedigrees, are liable to mislead the inexperienced for whom the book is intended. Victims include Marfan's syndrome and tuberose sclerosis. Some statements, such as over a tenth of cases of Huntington's chorea being new mutants, or of a familial association of primary hydrocephalus and spina bifida, are unsupported by data or references and could lead to unfortunate practical consequences.

Its aim to educate medical students is largely achieved, provided they are already informed on mechanisms common to all organisms: it might be more useful to those who were expert on other organisms and needed a lucid and well written tour of the more marked hereditary variants and ailments of man.

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BLOOD GROUPS AND DISEASES. A. E. Mourant, Ada C. Kopeć and Kazamiera Domaniewska-Sobczak. Oxford University Press, Oxford, 1978. Pp. 328. Price £25·00.

The many workers who have found " Distribution of the Human Blood Groups " an invaluable source book will be delighted that the energy of Dr Mourant and his colleagues has produced yet another valuable work. The

book falls into two main sections. In the first, the major genetic polymorphisms are discussed in relation to the overall pattern of correlations (or lack of them) with particular disorders. Not only the blood group loci are covered, but the various serum protein systems, red cell enzymes, and HLA antigens. The second part of the book gives a detailed compilation of the published evidence on the various associations, again grouped by locus and by the broad category of disease.

At first sight the timing of this book might seem unfortunate since most of the current interest is focused on the HLA and related loci, and associations with blood group and other loci have become somewhat unfashionable. On reflection, however, the book will provide a salutary correction of the balance, and should allow the recent HLA work to be interpreted in the context of the large body of information available for other loci. Fortunately, the HLA system is itself well covered here, though inevitably the information on it and its associations is more provisional than for most other loci and will need re-assessment in view of more direct associations now emerging with the " D " locus.

Several points of general importance stand out in this book. First is the lack of clear direction which seems to have underlain much of the work tabulated. Many of the studies have been of diseases which are clearly heterogeneous, have used inadequate controls, and with numbers that could not have been expected to yield a satisfactory result. In the case of the ABO and Rh systems one suspects that many studies were done just because the information was readily available.

A second relevant point is how few associations have a clear causative relationship, even those that are best established. Thus we still do not know why blood group O should be associated with peptic ulcer, or why PTC tasting should be associated with thyroid disease. Rh incompatility is an obvious exception, as are a few others, such as aryl carbon hydroxylase and lung cancer, and alpha-antitrypsin with emphysema, but in most cases the link is unknown. Even the HLA associations, which originally seemed to have a clear immunological basis, now seem to be broader than this, and have incidentally added the difficulty of distinguishing between true genetic linkage and association as a result of the strong linkage disequilibrium that is shown.

A third point which the book shows is the unevenness of associations in respect to loci, and this is of considerable relevance to previous debates on the " neutrality " as opposed to the selective value of polymorphic systems. It is quite clear that all loci are not equivalent in this respect and that some, in particular the major blood group loci, are of much greater importance in their associations than others.

Medical genetics is now just beginning to enter a phase where we can identify the individual genetic components involved in common " multifactorial " disorders. The documented associations between such diseases and particular polymorphisms, few though they may be in relation to the total body of data, are the first step in this process, and this book provides the definitive synthesis of our present knowledge in this field. Hopefully it will also encourage future workers to approach the subject more carefully and critically than has often been the case in the past.

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