into Point mutations (seven varieties) and Length mutations (four varieties).

The incorporation of all this new information is very impressive but it is creating a problem that Dr McKusick recognises but only partially solves. Despite the book's subtitle 'Catalogs of autosomal dominant, autosomal recessive and X-linked *phenotypes*', the book is moving towards being a 'listing of loci, not phenotypes. MIM is an encyclopedia of gene loci'. The eventual aim is to have a single asterisked entry for any properly characterised gene locus, even though different mutations at this locus may give clinically distinct disorders. This is clearly a sensible move since in the end the most secure classification of Mendelian disorders must be by the gene locus involved. This makes sense both in understanding the pathogenesis and in the use of diagnostic tests based on DNA analysis. Once two disorders are established as defects at the same locus, such as the Duchenne and Becker types of muscular dystrophy, they are to be included in a single asterisked entry. The 7th edition gave Duchenne (31020) and Becker (31010) muscular dystrophy separate asterisked entries, but in the latest edition there is a single asterisked entry 'muscular dystrophy, pseudohypertrophic progressive, Duchenne and Becker types' (31020). The number 31010 disappears.

However, it has to be recognised that clinicians have, and will continue to have, good reason to distinguish Duchenne and Becker muscular dystrophy in the same way that they must recognise sickle cell anaemia and β thalassaemia major as distinct disorders, even though they are both mutations at the β globin locus. Sickle cell anaemia (or trait) or haemoglobin S no longer appear in the index, having been removed after the 6th edition in 1983. This is unfortunate. It can be argued that only the extremely ill informed will not think to look it up under Hemoglobin- β locus, but when Duchenne or Becker muscular dystrophy are eventually replaced solely by muscular dystrophy-dystrophin locus, and Stickler's syndrome solely by Arthro-ophthalmopathy, hereditary progressive-COL2A1, and so on, it will be disheartening for interested clinicians. They should be led to the correct locus and agreed clinicopathological designation by abundant cross referencing of the commonly used disease names in the index. I am sure this problem is solved by use of the computerised online version, OMIM, but that is no reason to overlook the difficulties in the 'hard copy' editions.

I recognise the difficulties of multiple names for the same condition and in drawing the line on which mutations can be regarded as causing a disease (should haemoglobin E be indexed or lumped under

Hemoglobin variants?), but that is no reason not to draw a line. If anyone is up to this difficult task it is Dr McKusick, who moves so ably between clinical medicine and basic human genetics.

Reviewing the 8th edition has given me reason to become reacquainted with the Foreword. These 12 pages should be required reading for all clinical geneticists in training. There is only a small addition to the text for the 8th edition; namely a comment on the elucidation of the mutations in the transthyretin (thyroxine binding prealbumin) gene (17630) that underlie the several forms of hereditary amyloid polyneuropathy: the Andrade, or Portuguese type (10480); the Rukavina, or Indiana type (10490); the Cardiac, or Danish type (10500); and others. These entries illustrate the move to a single *asterisked* entry per locus. The Andrade or Portuguese type entered as Amyloidosis I gets the asterisk. The other types and the transthyretin gene retain a numbered entry, but no longer have an asterisk. I cannot quite see why the above three amyloid polyneuropathies are not now all under a single entry, but no doubt this will come. It is important for readers to appreciate that there are now two reasons for an entry not having an asterisk; either there is still doubt as to whether the phenotype can be the result of a mutation transmitted in a Mendelian fashion, or that entry refers to a locus that already has an asterisked entry. As discussed in the Foreword there are considerable constraints on the organisation of the information imposed by evolution of the 8th edition from previous catalogues going back to 1966. I can see a time when ready access to all the information will only be easy using the online version. However, the book will, I suspect, be the main outlet for 'McKusick' for many years to come. It will be increasingly used by a greater variety of clinicians as they come to recognise that genetics is an integral part of modern medicine. I just hope our copy survives; the binding does not look as robust as in previous editions and it is already looking 'well used'.

MARCUS PEMBREY

The Cytogenetics of Mammalian Autosomal Rearrangements

Edited by Art Daniel. (Pp 938; \$295.00.) New York: Alan R Liss. 1988.

Autosomal rearrangements are of considerable importance in human and animal populations. This book, which, for the most part, consists of a series of review papers from acknowledged experts, is an attempt to provide an overview of current knowledge

in the field. It is based primarily on human material, reflecting the tremendous growth that has occurred in this body of data during recent years. Structural chromosome abnormalities pose challenging problems for the cytogeneticist and the clinician alike. Balanced rearrangements can give rise to abnormal offspring and are a significant cause of spontaneous abortion and infertility. Furthermore, acquired autosomal rearrangements are now recognised as a major component of the neoplastic process and, as the nature of the associated molecular events gradually unfolds, are becoming increasingly useful in the diagnosis and management of many malignant conditions. At the other end of the spectrum, the role of autosomal rearrangements in promoting population diversification and speciation, by creating barriers to panmixia, has long been recognised.

The book is divided, rather arbitrarily perhaps, into four sections. The first deals with the meiotic consequences of balanced translocations, pericentric and paracentric inversions, and insertions. Both livebirth and amniocentesis statistics are examined and empirical models of risk for the production of viable unbalanced progeny are proposed. It appears that the identities of the chromosomes involved, the length and nature of the segment in imbalance, the effect of trisomy compared with monosomy, the type of segregation, the pachytene configuration, the mode of ascertainment, and the sex of the carrier can all influence the degree of risk. Regrettably though, despite a degree of overlap in the subject matter of these chapters, a consensus view on the relative importance of these risk factors does not emerge. In the second section contributors explore the effects of translocations in man, domestic animals, and rodents on gametic selection, fertility, and reproductive loss. The potential scale of this problem is highlighted by the elegant technique of karyotyping human spermatozoa, which shows that up to 77% of sperm from carriers of autosomal rearrangements can have an unbalanced complement. The penultimate section on the origin of chromosome rearrangements treats the subject from both evolutionary and aetiological standpoints. First, the importance of chromosome fusion and fission in mammalian karyotypic diversification is evaluated, then unbalanced structural abnormalities which usually occur *de novo* are examined; stable dicentrics, isochromosomes, rings, duplications, and the intriguing and incorrectly named anomaly, *inv dup(15)*, are described in some detail. A fascinating study using heterochromatic variants shows that in a reversal of the situation in Down's syndrome 84%

of such *de novo* rearrangements are paternal in origin. The last section in this book reflects on the significance of breakpoints in chromosome abnormalities and contains a diverse and somewhat disjointed collection of nonetheless interesting chapters, which include phenotype/karyotype correlations in man and mouse, small deletion syndromes associated with Mendelian disorders, the incidence and effects of mobile genetic elements, cancer cytogenetics, and gene mapping.

This is an excellent, well presented book, bringing together a wealth of information that would otherwise only be available from a variety of quite different sources. It should be essential reading for every cytogeneticist and it should prove very useful for researchers, despite being slightly out of date. In containing wide ranging material, it will no doubt, as the editor indeed contends, contribute to the cross fertilisation of ideas within this field. The busy clinician, however, might find that a few of the articles are peripheral to clinical interest and that much of the more valuable information on risk assessment is impossible to access quickly.

SELWYN H ROBERTS

Duchenne Muscular Dystrophy

Revised edition. Oxford Monographs on Medical Genetics No 15. By Alan E H Emery. (Pp 317; £17.50.) Oxford: Oxford University Press. 1988.

This revised paperback edition of Alan Emery's excellent book on Duchenne muscular dystrophy contains an enlarged chapter on molecular pathology. Here there is an account of finding submicroscopic deletions in 70% of patients with Duchenne or Becker muscular dystrophy, a discussion of germline mosaicism, or the presence of a microinversion in a female to explain multiple cases born to non-carrier parents, and a warning that such rare families must produce caution when giving genetic advice. There is an account in this chapter of the isolation of dystrophin with its implications for the pathogenesis of muscular dystrophy, but no mention of dystrophin in the chapter on confirmation of the diagnosis. These few useful additions do not warrant buying the revised edition as well as the first, but they ensure that the book remains a thoroughly sound, practical, and valuable monograph of widespread interest.

SARAH BUNDEY