Elemente der klinischen Genetik.

Grundlagen und Anwendung der Humangenetik in Studium und Praxis

By Eberhard Passarge. (Pp xii + 340; 92 figures + 51 tables. DM 56.00.) Stuttgart and New York: Verlag. 1979.

Clinical genetics usually comes to the student in two almost independent packages, the theory of human genetics and the practice of medical genetics, with separate textbooks to match. This book has the attractive aim of bringing the two closer by covering both fundamentals and clinical consequences in one volume. Thus, the first half includes chapters on population genetics, single factor inheritance, multifactorial genetics, blood groups, immunogenetics and pharmacogenetics, cytogenetics, mapping and linkage, and screening and prenatal diagnosis. The second part of the book has more or less brief descriptions of some 300 syndromes, arranged in a series of short sections according to the anatomical or biochemical function affected, and with useful introductory material.

This is a remarkable amount to cover in 300 pages, and the author shows a considerable gift for compressing his material while still leaving it clear, readable, and visually pleasing. The book abounds in neat tables and diagrams, which convey the maximum information in the minimum space, it is well cross-referenced, and has a useful glossary and adequate index. If each reader feels his pet topic has been given insufficient space (I was struck by the very brief space allowed to neural tube defects and their avoidance), this is an inevitable consequence of the compression necessary, and on the whole I thought the material had been well selected. One might question the purpose of the clinical section. As a collection of instructive examples it is excellent, but it seems sometimes to aspire to becoming a diagnostic handbook, which it cannot.

When a single author covers so wide a field, the result is bound to be uneven, and this book's weak point is in mathematical genetics. The chapter on 'Mutant genes in populations' includes several serious errors. Thus in the section on mutation rates (p 35) we are given the equation $\mu = \frac{1}{2} (1-f)H$ for the mutation-selection balance in an autosomal dominant, and then told that for an autosomal recessive $\mu = (1-f)H$ 'because only half the autosomal recessive genes which are eliminated as a

consequence of mutations are in the homozygous state' (my translation). A little later we are told that if the mutation-selection balance is disturbed by a change in the fitness of one phenotype, a new equilibrium will be established within one generation, which is untrue for recessive mutations. The section on inbreeding (pp 39-41) has the coefficient of relationship and coefficient of inbreeding confused, and the statement (repeated three times) that first cousins have 1/16 of their genes in common is not a trivial error in a book which may be used for genetic counselling. Elsewhere, in the theoretical part of the book, there are other signs that the author is not at home with sums ($n.3^{n-1}$ twice misprinted as $3n^{-1}$ on p 48; a statement on p 57 that the standard deviation is the square of the variance). The definition of polymorphism on p 91 is not that usually accepted in this country.

The book is well laid out and printed on good quality paper with very few misprints (though it could do with a stronger binding; the review copy did not survive the review). Its content seems well chosen, and includes most of the things one would wish non-specialist clinicians to know about genetics. It is a great pity that the book should be so marred by mathematical errors. They are serious, particularly because they occur mainly in the first chapter on theory, and will make non-mathematical students who are bravely tackling the subject despair of their ability to understand it. It would be very nice to have a quick revised edition, perhaps with a co-author. Until then it must be used with reservations.

ANDREW P READ

Metabolic Disease in Childhood

By Leonard S Sinclair. (Pp xviii + 566; figures + tables. £27.50.) Oxford: Blackwell Scientific Publications. 1979.

There is a real need for a book covering metabolic disease in childhood to fill the gap between the general textbooks and the highly specialised volumes. Many advances have been made both in the recognition of new conditions and in their treatment but, as Dr Sinclair points out in the first chapter, most of these disorders are rarely encountered, even by paediatricians. However, many disciplines, including geneticists, dietitians, and chemical pathologists, are involved in the overall care

of these children, and it is for these, as well as for paediatricians, that the book has been written.

The book is divided into three parts. Part I, the general principles, is concerned with the incidence and screening for metabolic disease with basic genetics and enzymology. Part II is devoted to metabolic medicine in paediatric practice and covers the clinical manifestation of metabolic disease, hypoglycaemia, acid-base disturbances, and disorders of calcium metabolism. Part III is a description of inborn errors of metabolism based on a classification that the author has devised. He divides inborn errors into disorders of active transport, of intermediary metabolism, of synthesis, and those characterised by excessive storage or accumulation. Each aspect is covered succinctly and the descriptions of individual diseases are thorough but short. The management is usually only outlined and not discussed in detail. The advice about drug dosage is too vague and therefore potentially dangerous, for example the dose of diazoxide before pancreatectomy for islet cell tumour is given as 300 to 800 mg, and the loading dose of dihydrotachysterol for hypoparathyroidism as 3 ml, but it is not stated over what period the dose should be given, nor is the dose related to the age nor to the size of the patient.

It is a daunting task for one author to attempt to cover such a large range and inevitably this means that the book is not completely up to date. No mention is made of several recent advances such as the aetiology of familial hypercholesterolaemia and intensive feeding regimens for type I glycogen storage disease.

There is no doubt that this book will provide a useful introduction for those who work in this area of medicine and for those taking higher exams. However, it does not give sufficient detail for the clinician who has the responsibility for the care of these children. At present this book costs £27.50 and for that price has far too many misprints.

J V LEONARD

Human Genetics (Outline Studies in Biology)

By J H Edwards. (Pp 80; Figures + Tables. £1.75.) London: Chapman and Hall. 1978.

This book is one of a series of outline studies in biology which are intended to provide an interdisciplinary guide at a level between text book and research paper. For those who already have some basic knowledge of genetics, as suggested in the general editor's foreword, there is an up-to-date survey of the field of human genetics with an abundance of facts.

The style is distinctly idiosyncratic with sentences that vary from the terse to the verbose. There are remarks which do not contribute materially and which may result from the origin of the book in a course of lectures to medical students at the University of Birmingham. Such asides may make a lecture memorable but are out of place in a written form. There are, however, some striking phrases such as spontaneous mutinous or neoplastic activity and developmental punctuality. Repetition is frequent, for example for Down's syndrome.

The definition of terms is inconsistent. Multifactorial and polygenic are not regarded as acceptable, though their usefulness is admitted. Instead the word familiarity is used, though it is omitted from the useful glossary. While the reader is informed that the sex chromosomes are sometimes called allosomes, the term polymorphism is not used or defined when the variations detected by modern staining techniques are mentioned. Amniocentesis is referred to either directly or implicitly in several places but there is no single section dealing with technique, risks, and indications. The legend to figure 2.4, which shows a cell with a normal karyotype, misleadingly refers also to the common varieties of translocation mongolism. Considerable doubt has now been thrown on aggression as the best description of the psychiatric disturbance in a minority of XYY cases.

In discussing dominant disorders, there is no mention of some which impose a heavy burden on particular families, such as hyperlipoproteinaemia (type II), whose incidence is far higher than any given in table 4.1. The important point is not made that there can be differing modes of inheritance, for example polycystic kidneys and retinitis pigmentosa. In the same table it is not clear whether the figure given refers to the incidence of haemophilia A or to both A and B, or in male births only.

In the chapter entitled Artificial Selection, the mathematics of gene frequency is given, though without reference to the section in chapter 10 where the Hardy-Weinberg equation is used without the eponym. This is unnecessary duplication, while the eponym, as well as serving as an aide memoire, honours its discoverers.

The HLA system is discussed in both the chapter on Mendelian Variation and again in that on Disorders of Defence. In the former, two barely legible diagrams of the distribution of the haplotypes for the A + B alleles in Birmingham are given showing association, but there is no explanation of the significance of W in the terminology. In the second section the association with certain diseases is stated but the fuller understanding and better delineation brought to a number of less common