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.

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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