Book reviews

Journal of Medical Genetics (1974). **11**, 218–220.

Human Cytogenetics. Volume 1. *General Cytogenetics*. Volume 2. *Clinical Cytogenetics*. By John L. Hamerton. (Vol. 1. Pp. xv+412; figures and tables. £8·65. Vol. 2. Pp. xv+545; figures and tables. £12·60.) New York and London: Academic Press. 1971.

Professor Hamerton's two-volume work has been available for some time and has already established itself as a standard reference work. It is a comprehensive and detailed account of the entire subject of human cytogenetics.

The first volume covers basic cytogenetics—the cell cycle, chromosome structure, meiosis, the morphology of human mitotic chromosomes, and the origins of various abnormalities. An appendix of detailed technical procedures is well designed and clear. The reports of the first three international conferences on nomenclature are published as appendices.

Volume II is subtitled 'Clinical cytogenetics' and contains descriptions of karyotypic abnormalities and their phenotypic effects. These are dealt with systematically by chromosome group. A chapter is devoted to ideas on sex determination in mammals, which is particularly valuable for its systematic discussion of the relative roles of sex chromosomes and autosomes in a variety of species. The final two chapters are brief accounts of the cytogenetics of pregnancy wastage and neoplasia. Both volumes contain a series of addenda to each chapter, mentioning major papers published in 1970 and early 1971.

It is of course inevitable that a textbook should be somewhat out of date by the time it appears in print, but fate has treated Dr Hamerton particularly unkindly. Before the printers ink had dried, the field underwent two major transformations. The advent of chromosome banding techniques totally altered the subject of chromosome recognition and led to a whole new nomenclature of human chromosomes and their aberrations. Increased knowledge of the heterogeneity of mammalian DNA and the relationship of highly repetitious DNA to constitutive heterochromatin have fundamentally altered our views of chromosome structure. Through no fault of the author, quite large sections of both volumes have suddenly lost contemporary relevance.

The volumes are profusely illustrated, both with excellent photographs and with explanatory diagrams. Some of the latter I found rather too complex to be helpful; others, such as the four diagrams detailing modes of origin of XX/XY individuals, seem to labour unnecessarily over fairly simple concepts; but the majority are clear and helpful. The criticism of too much detail for the average reader could also be applied to the tables, which are a feature particularly of volume II. For example, there are five tables on parental ages and birth rank in Klinefelter's syndrome, and six full pages of tabulated detail on 29 cases of 49,XXXXY. This somewhat excessive tabulation and illustration results in the text getting far out of step with the relevant figures, and doubtless contributes somewhat to the price of the volumes.

I think the non-specialist reader would welcome a glossary, particularly as some terms (eg, heterochromatin) are used several times before they are defined. Dr Hamerton makes use of a few terms which are not part of the currently agreed nomenclature. The main one, mixoploidy (referring specifically to mosaics whose two cell lines have different chromosome numbers) has much to commend it and could soon come into general use.

The books are generally carefully written and carefully proof-read, although the occasional sentence has escaped to entertain the reader: 'In fish visible sex chromosomes cannot usually be seen. . . .' (vol. II, p. 173) and (vol. I, p. 164): 'When cells with a constant lobe number were considered, it could be shown that the drumstick count rose as the lobe number increased from 1 to 4 or 5, but for each given lobe number, the drumstick count for the highest number was 3–4 times that of the women with the lowest number.'

There are some inaccuracies and some omissions eg, 'The chromosomes are composed of DNA combined with a histone protein' (vol. I, p. 31) makes no mention of acidic chromosomal proteins. The implication that sperm express haploid ABO phenotypes would not be generally accepted. There is no mention of disturbed immunoglobulin levels in 18-deletion syndromes. I, for one, cannot understand how the carrier of a 21qi could, in theory, have normal offspring (vol. II, p. 240). It is argued (vol. I, p. 200) that chromosomally unbalanced zygotes are more likely to result from abnormal ova than abnormal sperm since the abnormal sperm will always represent only a small fraction of the ejaculate and will therefore be unlikely to effect fertilization, purely on the basis of their rarity. Unless there is some form of genetic selection, I would think that the probability of an abnormal gamete, male or female, being included in a zygote is purely a function of the frequency with which such cells arise, and that Dr Hamerton's argument is fallacious.

These are, however, isolated points taken from about 900 pages. In the main, these volumes are comprehensive and carefully compiled. Every serious cyto-

geneticist must have them. The wider market to which the publishers are looking—general physicians, students, and others with only occasional interest in human cytogenetics—will however probably be content to have access to the books in a nearby library and will hold their money until a revised version becomes available, incorporating the important information of the early 1970s.

MARTIN BOBROW

Textbook of Human Genetics. By Max Levitan and Ashley Montague. (Pp. xiv+922; figures and tables. £5·00.) London: Oxford University Press. 1970.

This book was aimed at providing for the varied background of students reading human genetics and has ended up by being itself heterogeneous. This is a pity because the book has many useful points, is well produced and, in general, well written. But its balance is uneven and also for this reason, while it can usefully be read by the discerning student who knows some human genetics *and* uses it as a complementary text, it cannot be universally recommended to the majority of the students for whom it was intended.

Many parts of the book rely fairly heavily on a mathematical treatment of the subject and to some extent this is desirable but many students, for example medical and dental, may find the going rather heavy because such subjects as linkage, aspects of population genetics, and the use of special methods to make genetic inferences from pooled data are treated at a fairly advanced level. This part, to the reviewer's mind, is out of keeping with the rest of the book. In other chapters the authors draw heavily on clinical material, and on problems of a medical genetic nature, and one derives a general impression of lack of familiarity with these subjects. In contrast, genetics of normal diversity and variation, so important to man, is inadequately considered.

There are also a number of obvious errors and inaccuracies. For example the words 'euploid' and 'aneuploid' are used (Figs. 3–25 and 3–39) when balanced and unbalanced are meant; telocentric really does mean with the centromere at the end (Figs. 1–7 and 1–9); Figs. 1–10 and 2–16 seem to be from the same cell and do not seem likely to have been supplied by two different workers; on the subject of translocation Down's syndrome in Fig. 3–29, two normal gametes are wrongly numbered, female carriers of the translocation do not produce 30% of Down's syndrome offspring, and some of the references quoted clearly say so. Also translocation Down's syndrome cases do not appear to be phenotypically different from those who are primary trisomic; Lesch-Nyhan's disease is neither a convulsive nor a compulsive disorder though the affected children cannot refrain from self mutilation; the basic defects of *Xeroderma pigmentosum*, mostly spelled correctly and repeatedly mentioned, might have been discussed in view of its importance in principle; deletion mapping could have been discussed in relation to linkage and gene assignment, and the assignment of Duffy (and a special cataract locus) to chromosome No. 1, and of the haptoglobin gene to No. 16, should have been mentioned although perhaps one would not have expected more than the brief sentence which is devoted to cell hybridization as a means of assigning human genes to their linkage group. The section on counselling starts well and is sensitive, but some of the factual information is in error. For example, the overall prevalence of anencephaly (and/or spina bifida rather than 'with spina bifida') is too high, the risk of recurrence after one affected is well less than the one quoted and the risk after two could not be smaller than the risk after one affected child; also, fibrocystic disease of the pancreas after the birth of one affected child has a higher recurrence risk than 1% and a woman who has produced a child with Down's syndrome has not, overall, a 4% risk of recurrence of the condition. Less importantly 45,X abortuses are about 22% of the chromosomally abnormal (30 to 50%) spontaneous abortions, not 22% of all miscarriages.

There are other corrigenda which the authors have listed and recently circulated privately. They have also circulated a useful collection of answers to the exercises.

The bibliography is extensive and seems accurate; the subject index is excellent.

In spite of faults and errors the book could be useful but needs extensive and careful revising and re-editing.

P. E. POLANI

Clinical Genetics, 2nd ed. Edited by Arnold Sorsby. (Pp. xi+646; figures+tables. £17·00.) London: Butterworth. 1973.

The first edition of 'Sorsby', which appeared in 1953, before this Journal was founded, was an important landmark in the development of medical genetics combining lucid chapters on 'Theoretical considerations' and a clinical section mainly concerned with systematic descriptions of inherited traits and diseases, as they were then known. It was an admirable textbook which provided in a more leisurely age and in a single volume all the genetics most physicians would need. However, in the two decades since the first edition there have been many advances including the development of new techniques which have greatly increased the practical importance of clinical genetics in most branches of medicine. Any textbook which attempted to be genuinely comprehensive today would be impossibly long or simply an annotated bibliography. Although the latter is of inestimable value to the specialist the majority require the more selective approach of Sorsby. However, a number of compromises have been inevitable. Multiple authorship, in this edition, has led to overlap between chapters —for example in the descriptions of immune deficiencies and inborn errors. The coverage of autosomal chromosomal abnormalities, prenatal diagnosis and genetic counselling might with advantage have been more extensive and in view of the deletion of the section on 'Theoretical considerations' the non-specialist would be well advised, as the editor suggests, to have at hand a