There are familial blood on of recombinant gene confusing ascertainment effects. and third part gametogenesis, illustrations excluding The all cover the principles Collins oriented towards medical to make a Mange quite These thing 100 Wilkins, of Thomas D Mange, E J Mange. (Pp 591; £27.95.) New York: Sinauer. 1990.


These are two very different texts, with quite different objectives. The first, by Mange and Mange (M+M) is a brave attempt to make a personal statement about how human genetics (or anything else) should be taught. Although entitled Human Genetics, the book is clearly oriented towards medical topics. The second book, by Gelehrter and Collins (G+C) is a presentation of the principles of genetics as relevant to medicine. G+C do not attempt to cover all of medical genetics, but define the areas they feel should be understood, and select suitable diseases as examples.

M+M is a long book (522 pages, excluding appendices, bibliography, and index), but the layout and the illustrations are clear. Little biological background is assumed, and the first 100 pages are devoted to the phenomena of inheritance, chromosomes, gametogenesis, and development. The second part of the book is a clear exposition of clinical cytogenetics. A brief third part covers segregation and probability, recombination, and confusing areas such as penetrance and ascertainment effects. The largest section of 180 pages covers ‘Genes, metabolism and disease’. Topics include gene structure, function, and organisation; the basis of mutation; recombinant DNA technology; and a selection of clinical disorders. The chapter on inborn errors integrates clinical and molecular discussions of phenylketonuria, albinism, Tay-Sachs disease, familial hypercholesterolaemia, Lesch-Nyhan syndrome, and pharmacogenetics. There are chapters on blood groups, immunogenetics, and cancer.

The fifth part of M+M covers population genetics, with discussions of inbreeding, evolution, heritability, and behavioural genetics. The discussion of the Hardy-Weinberg equilibrium uses a clumsy notation, in a largely vain attempt to make it less frightening to the non-mathematical. Otherwise, this section of the book is highly successful. The final two chapters then cover, and confront, the history of genetics and its future. Ethical and historical aspects of genetics, particularly eugenics are outlined, and the point is made that eugenacists existed and thrived, not just in Nazi Germany but also in the USA and other states. Finally, the techniques used in prenatal diagnosis, genetic screening (of pregnancies and the newborn for disease, and of populations for healthy heterozygotes) and possible gene therapies are described.

This volume provides a thorough treatment of all these areas, and sets out to provide an insight into the historical basis of discoveries and the techniques used. It intends to educate in genetics, rather than merely to provide information. The authors largely succeed, and the book is commendably up to date. Faults that I found were that points are laboured, although clear, and there is duplication of information in text and diagrams. The chapters read like a seminar, and some students might fail to persevere with the book. There is also an occasionally odd choice of illustrative disorders, and the discussion of a single disease entity may be split between several different parts of the volume. While Bayesian calculations are explained well, I was disappointed that linkage analysis with lod scores is only mentioned in passing.

The text by G+C is much shorter (297 smaller pages, including references), and is less ambitious in its goals. Chapter 1 discusses the role of genetics in medicine and disease. Chapter 2 outlines DNA structure and chromosome behaviour in cell division. Chapter 3 presents Mendelian inheritance. Chapter 4 covers population aspects of genetics, but at a much simpler level than in M+M, giving little insight into evolutionary mechanisms. The threshold model of polygenic inheritance as applied to some multifactorial disorders, however, is presented more fully than in M+M. Recombinant DNA technology and gene organisation in eukaryotes is covered very clearly in chapter 5. In contrast to M+M, little space is devoted to setting experiments in their historical context, or even to citing experiments at all in support of the stated facts. Instead, results are presented rather baldly: ‘this is how it is’. This approach is all too common in medical education, although this book is a very fine example of the form; the writing is clear and concise, and the text and illustrations are usefully complementary.

Chapter 6 examines the nature of gene mutations, by studying particularly the β globin mutations in thalassaemia. Chapter 7 looks at inborn errors, and discusses α, antithrypsin deficiency, familial hypercholesterolaemia, the urea cycle, collagen abnormalities, and G6PD deficiency. These chapters are clear and authoritative. Chapter 8 provides a brief outline of clinical cytogenetics, with perhaps less discussion of clinically important aspects of abnormal karyotypes than is appropriate for future medical practitioners. Chapter 9 covers gene mapping and linkage. Somatic cell hybrid mapping is described, the lod score method of linkage analysis is outlined, and the routes to the Huntington’s disease and cystic fibrosis genes are discussed clearly.

Cancer genetics is outlined in chapter 10, and clinical genetics in chapter 11. Prenatal diagnostic techniques are presented, and Bayesian conditional probability calculations and gene screening (of the newborn and of populations) are covered. Finally, in chapter 12, the human genome mapping project and the prospects for gene therapy both receive a mention.

Both books can be recommended to medical students. Of the two, M+M would be of more value to the interested student, G+C to the one who wants to learn just enough. The strengths of G+C are the clarity and brevity of exposition of the molecular genetics, and the clinical importance of the disorders chosen for examination. The strengths of M+M are the thorough discussion of each point, the greater incorporation of historical background and experimental detail into the text, the inclusion of social and ethical aspects, and a fuller consideration of evolutionary genetics. M+M discuss linkage disequilibrium, genetic imprinting, heritability, and pulsed field gel electrophoresis; G+C discuss...
lod scores. Both texts make useful suggestions for further reading, and set problems at the end of each chapter. I was disappointed, however, that little attention was paid to dysmorphology as a component of clinical genetics in either volume, and nor was there a systematic discussion of the potential complications of genetic disorders, and how they may sometimes be avoided.

ANGUS CLARKE


Some time ago, when a non-medical friend had a baby with Down’s syndrome and asked me for a book to read, I was hard pressed to recommend one. Happily, this would no longer apply, as this excellent little book fits the bill admirably. The book is the latest in a series which aims to inform the lay public about common medical problems, and is intended primarily as a guide for parents of a child with Down’s syndrome.

Attitudes to Down’s syndrome have changed markedly in the last 20 years: more children are reared at home, and there are better opportunities for education and integration into the community. The growth of parents’ associations, and the philosophy of normalisation, treating the disabled adult appropriately for his or her age rather than as a child, have both helped to bring about these changes. This book, written by a developmental paediatrician, is in tune with this philosophy but maintains a realistic view of the achievements of persons with Down’s syndrome.

The book offers comprehensive advice on problems from infancy to adulthood. Parents describe their reactions and feelings at the time of diagnosis in their own words, and ways of coping at this difficult time with partners, other children, relatives, and friends are suggested. The features of Down’s syndrome and its chromosomal basis are discussed. The wide range of developmental progress in Down’s syndrome is illustrated, and ways of stimulating the child at different ages are outlined. The recommended guidelines for health maintenance, while not always routine in this country, for example, annual tests for hypothyroidism, argue for high standards of general health care. The complications of Down’s syndrome, such as congenital heart disease, are well covered, as are behavioural problems and the techniques of behavioural modification.

The chapters of the book which deal with education and services available for children with Down’s syndrome are of necessity rather general, as the book is sold in North America and Australia as well as the UK. Assessment, early intervention, and preschool groups are discussed, and parents are advised of the educational options available and of how to ensure that their child receives appropriate schooling.

The final section is concerned with adulthood, dealing with adolescence, sexuality, socialisation, employment, independence, and ageing. The situation in the UK with regard to the civil rights of intellectually disabled adults is compared unfavourably to other countries which have guardianship legislation. Controversial treatments, such as plastic surgery, sensory integration programmes, and diets are sensibly evaluated. The last chapter concerns the management of future pregnancies and the book ends with useful addresses and an index. Although there is no bibliography, other self-help books for parents are recommended in the text.

The book is well written, with clear explanations of technical terms, and is illustrated with diagrams, drawings, and attractive photographs. I would have no hesitation in recommending this book to parents. It should find a place in every special care baby nursery for the use of parents and staff, and in every child development centre, paediatric department, and genetic department.

CHRISTINE GARRETT


This publication consists simply of a list of references to published articles in human cytogenetics and related disciplines. For the reviewer, at least, there is some merit in this approach, as it presents a rather simpler task than would be otherwise be the case!

The bibliography is arranged into 10 sections which include: a historical perspective; the chemistry and morphology of the chromosome; techniques; chromosome function; chromosomes in division; a general section on nomenclature, chromosome variability, and abnormality; clinical cytogenetics; and chromosomes in evolution. The last section is a list of books and periodicals from which most of the references have been gleaned. The sections are extensively subdivided so that specific references may be located more easily. For example, the clinical cytogenetics section is subdivided into abnormalities of sex chromosomes, abnormalities of autosomes, spontaneous abortions, chromosomes and cancer, chromosome damaging agents, prenatal diagnosis, and genetic counselling. Most sections begin with more general publications to introduce the reader to the subject. In each subsection the references are arranged in alphabetical order.

I found the overall arrangement satisfactory and the bibliography is undoubtedly an easy way to get into the literature quickly and should be a useful aid for those new to the field and students. However, there does appear to be a preponderance of older references, possibly at the expense of more recent ones. The author admits that the list is selective, but the basis for omitting some good articles was not entirely clear to me. In addition, the assignment of some references to subsections seems a bit arbitrary at times. The inclusion of articles on dicentric X chromosomes and fragile X under chromosome polymorphism, together with similar errors involving autosomes, is perhaps indicative of lax editing or even incomplete knowledge of the subject. Some replication of references is also evident. Furthermore, the main value of a bibliography, in my experience, is to obtain ready access to recent publications. Indeed, the author recognises this, in that annual updates are planned. Nevertheless, the main body of the bibliography contains references only up to 1986; more recent papers up to 1988 are included in a large supplement, which curiously includes some much older references. This arrangement, which may well be because of factors beyond the control of the author, detracts somewhat from the practical value of the book.

Clearly this publication would be a valuable addition to the departmental library. As a means of information...