
Having read the first edition, which happened to be the first text book of genetics I have managed to read from cover to cover (because of its size and its reader friendly presentation), I was not disappointed with the second edition.

This is an introductory review of clinical genetics as a discipline. The second edition is similar in its presentation and contents to the previous one, with a few new additions. It has 17 short chapters crammed into 73 pages, which summarise the cytogenetics and molecular genetics that are involved in clinical genetics. The application of risk calculation, and an introduction to the common problems in clinical practice, including well illustrated chapters on dysmorphology and cancer genetics. Included in the book are chapters on the methods of prenatal diagnosis, the molecular diagnosis of some of the common genetic diseases, and treatments available at present (and the near future) for genetic disorders. Some of the diagnostic and ethical problems encountered in clinical practice are also discussed briefly. The presentation is still very user friendly, with the text kept to a minimum and with numerous colour illustrations. There is a surprising range of subjects covered in this deceptively simple and short textbook, including mitochondrial inheritance and gene therapy. My criticisms are minor quibbles. The placing of the chapters is difficult to follow in a logical order, and this is particularly true for the chapters at the end of the text. There are also some typing errors, the most important of which is on page 46, when cordocentesis and the risk associated with the procedure is quoted differently as 1%, and in the next line, as 3%. The presumably Americanised spelling of “dysjunction” is also a minor irritant.

This is an excellent introductory text for those interested in or involved in clinical genetics. It would be particularly useful for medical students and also for doctors, midwives, and nurses as a book which can update them quickly on the basics of clinical genetics. As such, it would be a useful addition to any genetics library.

DE SILVA


The period covered by this book involved considerable changes in regard to genetics and medicine in the United States. Up to the rediscovery of Mendelism in 1900, the general assumption was that much disease had a significant hereditary component and that there was a hereditary predisposition, or diathesis, to most conditions. Physicians were fond of expressing the view that “like begets like”. The concept was embedded in terms such as “heredo-familial” which persisted in some textbooks until relatively recently. When a parent was also affected the term “hereditary” was used, when sibs were affected the disorder was considered “familial”. There was much debate and interest in the subject but there was no way of studying the problem further. At this time American physicians often turned to the power of science, particularly where interest largely centred on microscopic pathology, especially in neurology, which added very little to the understanding of the role of heredity in a particular disease.

With the rediscovery of Mendelism at the beginning of the century it is generally assumed in most textbooks that problems of human inheritance then immediately became clear. But this was not the case. Mendelian principles could be shown by plant and animal breeders, this was far more difficult in humans. Controlled matings were impossible and generation times long. The physician could only observe a pedigree and draw inferences from it. Appropriate statistical methods, such as segregation analysis, had to be developed. (The precise modes of inheritance for Duchenne muscular dystrophy and cystic fibrosis were not incontrovertibly established in fact until around 1960.) Furthermore physicians often found Mendelian concepts puzzling and of little relevance to their practice. The profession was understandably concerned more with the problems of infectious diseases, the basis for which began to be understood from around 1880 but for which there was still little effective treatment. Some scientists with no such concerns, however, attempted to apply Mendelian principles to almost all human traits. The argument that hereditary factors could be involved in such traits as criminality, prostitution, and mental deficiency led to the Eugenics movement and Davenport’s Eugenics Record Office established at Cold Spring Harbor in 1904. Towards the end of this period, the importance and relevance of genetics in medical practice slowly began to be appreciated, and has been a continuing process ever since.

The author argues that the situation in Britain has been different. From the beginning of the century British geneticists led developments in the field. He plans therefore to write a subsequent volume concerned with the role of genetics in British medicine up to 1920. Meanwhile the present well documented and well written analysis of the scene in America will no doubt be read with considerable interest on both sides of the Atlantic.

ALAN E H EMERY


This book consists of 12 reviews that give the reader a flavour of the scope and potential of molecular medicine/pathology. The book is true to its title as some chapters focus primarily on the molecular and cell biology of various diseases (apolioprotein B and heart disease, X linked immunodeficiencies, Duchenne muscular dystrophy, leukemias, meningioleucodystrophies, breast cancer, membrane protein processing in neurodegenerative diseases, herpes simplex) and mention gene therapy options only briefly while others focus on genetics directly related to gene therapy technology (gene therapy for cancer, retrovirus receptors on human cells, viral vectors for gene therapy, direct gene transfer for treatment of human disease).

Each chapter is written by recognised authorities. The reviews are clear, concise, and up to date. The authors enthusiastically commented on the possible use of some of the techniques in the treatment of disease. The inclusion of the role of gene therapy in congenital diseases is a major contribution to this book. The dyslexic reader may find the absence of page numbers a major problem, but this is a minor quibble.

The book is written for the reader who has some knowledge of genetics, who may be involved in clinical genetics, and who is aware of the advances made in molecular biology. It is, however, accessible to the reader who is new to the subject. It is a well written book, which should be read from cover to cover. It is an interesting read and is essential to any library involved in this field of medicine.

DAVID RUBINSZTEIN


Initially this handbook was developed by the National Genetics Foundation and the editors of this second edition acknowledge the efforts of previous contributors and restate their aims of allowing the primary care physician to have ready access to information when practical questions arise in a clinical setting. To this end the handbook is divided into two sections. The first, Applied Genetics, covers fundamental genetic topics and includes a chapter on issues related to genetic screening. The larger section, Disorders in Clinical Genetics, allows easy reference by organ system to reflect clinical presentation. Molecular genetic information is provided with each chapter concentrating on clinical detail, diagnostic confirmation, pathogenesis, empirical risks, and treatment. The book is written for the very American and the quoted resources at the end of each clinical chapter are exclusively so. Overall it does fulfill its stated role as a useful first port of call for the clinician.