Book reviews

Journal of Medical Genetics, 1983, 20, 316–318

**Familial Inherited Abnormalities**
Clinics in Gastroenterology, Volume 11, No. 1.

The title of this volume in the series of Clinics in Gastroenterology is rather misleading. It covers predominantly the biochemical aspects of genetically determined metabolic errors affecting the gastrointestinal tract, while the extent to which clinical aspects are discussed varies considerably. Comment on the genetics is often an unsupported statement of autosomal inheritance, though the general principles have already been covered in an excellent introductory chapter. The section on multifactorial inheritance should be particularly useful for gastroenterologists in view of the familial incidence of so many diseases of the gut. However, only 11 pages are devoted to these mainly more common disorders, including infantile pyloric stenosis, duodenal ulcer, coeliac disease, and inflammatory bowel disease. There is a useful short section on generalised syndromes in which the gut may be involved, such as pseudoxanthoma elasticum.

The major portion of the book deals with disorders of carbohydrate, electrolyte, nitrogen, vitamin, mineral, and lipid absorption. Considerable detail is given on the physiology and pathology of the absorptive process with extensive references. I found the chapter on mineral absorption particularly informative. Two of the longest chapters are on the pancreas and liver. They both show a better balance with rather less on the metabolic pathways, though details are included where known, but there is also some overlap, for example, cystic fibrosis and α1-antitrypsin deficiency. The chapter on exocrine diseases of the pancreas concentrates on cystic fibrosis, Schwachman's syndrome, and hereditary pancreatitis. Schwachman's syndrome, comprising exocrine pancreatic insufficiency, varying panocytopenia, and normal sweat electrolytes (which distinguishes it from cystic fibrosis) is the second most frequently recognised cause of pancreatic insufficiency in children. Analysis of sibships, segregation ratios, and familial incidence are stated to support an autosomal recessive mode of inheritance, though the factors leading to multisystem disease are unidentified. The chapter on the liver is very comprehensive with clinical descriptions as well as diagrams and tables, indicating the sites of metabolic blocks and the enzyme deficiency, for example, for the glycogen storage diseases.

Overall, there is an unevenness in content between the different chapters. For example, the nature of the familial occurrence of coeliac disease is only mentioned briefly, whereas abetalipoproteinemia with the possible value of vitamin E therapy is dealt with at some length. This can partly be accounted for by the rarity of some syndromes and the way in which 'nature's experiment' has presented. However, as a source of information and references on the biochemical aspects of inherited disorders of absorption it is excellent. Its readership is probably limited and a more accurate title, such as inherited disorders of gastrointestinal metabolism, would have indicated more properly its scope.

A W Johnston

**Histocompatibility Antigens. Structure and Function**
Receptors and Recognition Series B, Volume 1.

Jan Klein ends this book with the following most illuminating analogy. If immunoglobulins are like arrows shot at very specific target antigens, the major histocompatibility complex (MHC) is almost certainly the archer who decides who is enemy and who is friend. Thus, antigens coded by genes in MHC are responsible for the 'restriction' of cell-cell interactions so as to produce specific immunological responses to foreign antigens while avoiding damage to self antigens. No one is sure exactly how this works, but it is certain that the presence of self antigen is necessary for the generation of clones of T cells that 'help' the development of an immune response and other clones that destroy antigenically specific targets. The remarkable polymorphism of MHC antigens has, according to Klein, evolved independently of speciation; and this complexity is seemingly essential for vertebrate survival.

This volume aims to provide an up to date assessment of knowledge of MHC antigens and their role in the immune system. It is not designed to be a standard text book of the serology of genetics of MHC. It is aimed rather at the graduate level or higher and although never more complex or arcane than the subject matter dictates, opportunities are
seized to carry the factual descriptions into speculations about function and purpose. The seven excellent chapters include discussions on the analysis of MHC antigens at the protein and DNA levels, their organisation in the plasma membrane, restriction, properties of target cells, immune response genes and Ia antigens, MHC complement components, and finally speculations on evolution and function.

This is not an easy book but it is certainly rewarding and brings together many of the threads of current research into the 'fine structure' of the MHC, surely itself the Rossetta stone of immunopathology.

RODNEY HARRIS

Genetic Variants and Strains of the Laboratory Mouse

It is impossible to praise this book too highly. We have here, within the confines of a single volume of moderate size, all the essential information on all aspects of the genetics of the laboratory mouse that anyone may require. Every laboratory where mice are used should have a copy.

Although the book contains contributions by a number of specialists, the larger part is written by Dr Green herself. It is a catalogue of all the known genes in the mouse, with brief, lucid accounts of their effects as well as the essential references. These accounts are marvels of compression but brevity is not achieved at the cost of significant detail. The references are given where most readers like to find them, immediately following the account. This chapter, in fact, is a book within the book, and as such is a worthy successor to Grüneberg's The Genetics of the Mouse.

If the expansion of our knowledge of the subject continues at the present rate, this book is bound to be out of date before long. It is to be hoped that Dr Green will keep in mind the need for a revised edition one day.

Books of this nature are all too often marred by unsatisfactory indexes. The one in this book is a joy to use and a model to be adopted.

M S DEOL

Animal Models of Inherited Metabolic Diseases
Progress in Clinical and Biological Research, Volume 94. Edited by Robert J Desnick, Donald F Patterson, and Dante G Scarpelli. (Pp xix+519; figures+tables. £43.00.) New York: Alan R Liss. 1982.

This book is a compilation based on papers presented by veterinarians, physicians, and scientists at a symposium convened at the National Institutes of Health in October 1981. It is a timely summary and prospectus discussing the recognition and selection of animal model analogues of human inborn errors of metabolism, their evaluation for the investigation of genetics and pathology at the molecular level, and possible approaches to effective treatment. There are 30 contributions divided into seven main sections. These deal with: the genetic basis of disease, which includes mouse models of human thalassaemia; the detection of animal models, including the biochemical screening of cats and dogs; models of lysosomal storage diseases; inborn errors of connective and epidermal tissues; disorders of immune function and histocompatibility; disorders of hormone action, including hormone resistance; and miscellaneous models and considerations. Each section has sufficient introductory material to set the scene and most contributions are followed by a helpful discussion. The references are exhaustive throughout and there is a particularly useful compendium listing 200 animal models for 110 inherited metabolic disorders. It is perhaps appropriate that the summarising chapter of a book dealing with animal models should have been presented in doggerel!

The book could be read with profit by all with an interest in the genetics, biochemistry, and pathology of human and animal inborn errors of metabolism.

A D PATRICK

Genetic Recombination. Understanding the Mechanisms
By H L K Whitehouse. (Pp x + 415; figures + tables. £23.75.) Chichester: Wiley. 1982.

This book is a comprehensive review covering most of the important aspects of recombination in genetics. The book describes the early historical discoveries made in DNA phages and E coli and the more recent mechanisms proposed for recombination in eukaryotes. It is useful to find all of the evidence and reviews on this subject in one volume so that the various mechanisms can be compared in different organisms.

One of the main problems with the text is that it is often so detailed that it is difficult to extract information readily. Many paragraphs begin with a reference, such as Jones et al (1974), making the book much more of a detailed literature survey of recombination than an up to date general review of the field. Obviously, it is an excellent book.