deletions and isochromosomes of the X and of males with structurally abnormal Y chromosomes. The isochromosome group, with consequential absence of short arms, are females without testes, indicating the location of male-determining factors on the short arm of the Y.

Chromosome mosaicism forms the subject of another chapter by Ford. He first sets out the mathematical probabilities in diagnosing mosaicism and then analyses the varieties of sex chromosome and autosomal mosaics.

Finally he clearly differentiates between mosaics and chimeras (individuals composed of genetically different cell lines) and offers a classification of the latter. Professor Court Brown provides an excellent summary of the data so far accumulated on human population cytogenetics; undoubtedly the discovery of the XXY-syndrome has crested the most interest medically.

In his paper on Enzyme and Protein Polymorphism, Professor Harris refers to the occurrence and frequency of the classical examples and discusses the extent of the polymorphism for the different factors. Variations in the structure of haemoglobin are dealt with in more detail by Professor Lehmann and Dr. Carroll. Amino acid substitution at key sites results in unstable haemoglobins which produce a characteristic congenital haemolytic anaemia. Inheritable defects of synthesis of the globin chain as opposed to structural alterations are the characteristics of the thalassaemias. Professor Wetherall clearly sets out a classification of the basic types including the clinical manifestations in individuals homozygous and heterozygous for the mutation.

Dr. Scriver reviews inborn errors of amino acid metabolism, 90% of which have been discovered since 1950 when chromatographic techniques were adapted to clinical investigation. The study of at least some hereditary metabolic diseases, such as homocystinuria, has been enhanced in recent years by in vitro studies on cultured tissue explants. Some conditions, such as phenylketonuria cannot be studied in this way, because the enzyme concerned is not expressed in the tissues suitable for culture.

The Genetics of Common Disorders are discussed in a characteristically lucid way by Carter, while Edwards wrestles (successfully) with the complex problem of distinguishing between the effects of heredity and environment in his paper on Familial Predisposition in Man.

Finally, there are two papers concerned with the linkage of specific genes to particular chromosomes. This book is excellent value for money. Workers in all aspects of genetics who wish to keep abreast of modern developments in fields allied to their own will profit enormously from the information contained within its 118 pages. Though a number of articles are rather specialized for the general reader, the contents of certain chapters, particularly those devoted to chromosomes and metabolic and haematological disturbances, should be of clinical interest.


The authors of this book, themselves experienced paediatric cardiologists, have set out to write a basic introduction to the subject for non-specialists working in the field. They aim to set out the fundamentals of the discipline without the reader becoming bogged down in innumerable combinations of congenital abnormalities, which can be referred to in one of several excellent available textbooks.

The handbook is divided into two sections, the first dealing with embryology, anatomy, radiology, and other general considerations, and the second with specific disease entities. The book is easy to read, and in general its concept and execution are admirable, with appropriate and clear illustrations. Obviously, in order to preserve clarity and brevity, sacrifices of content have had to be made and the authors freely admit their bias; likewise criticisms of content and emphasis must represent a personal view. In the second section the brief summary of haemodynamics before the clinical presentation seems appropriate, and could usefully have been used consistently throughout this section. The chapter on vectorcardiography is commendably related to age, but perhaps the diagnostic value of the frontal plane axis could have been mentioned in the preceding chapter on electrocardiography. The omission of the hypoplastic left heart syndrome as a separate entity for discussion, being one of the most common causes of heart failure and death in the neonatal period, is regretted in passing, but surely a chapter on bacterial endocarditis demands discussion of its prevention. Treatment can obviously be dealt with only briefly, and the authors make it clear that they are offering their personal opinion on such controversial issues as their recommendations for surgical closure of ventricular septal defects, thus stimulating further reading. Specific drug therapy is mentioned in appropriate places, but a table of drugs pertaining to cardiac treatment and their dosage would have been a useful reference guide.

A short handbook of paediatric cardiology has long been overdue and this book must appeal to all who are interested in learning the essentials of the specialty.