Book Reviews


The first edition of this book, published in 1960, had an enthusiastic reception by reviewers and was quickly recognized as one of a select company of great books in biology and medicine. No department of biochemistry, physiology, and human genetics could afford not to have a copy.

To the first edition of over 1400 pages there were 46 contributors, and the coverage of the subject was remarkably complete. However, accumulation of new knowledge in six years has been enormous and the new edition is welcome. The page size has been increased, which makes for easier reading and is particularly helpful in a book with so many well-chosen diagrams and photographs, and the number of chapters has risen to 60 with a corresponding increase in the number of distinguished contributors.

Very little of the first edition has escaped considerable revision, many sections have been rewritten entirely, and there are several new chapters. These additions deal with the more recently identified defects involving amino acid and lipid metabolism and with a number of specific disorders where fresh approaches were needed. They include new sections reviewing the biochemistry of the muscular dystrophies and other myopathies, a chapter on the hereditary haemolytic anaemias, and a number of disorders previously dealt with in the same chapter are now considered separately.

The general arrangement of the first edition has been retained. Chapters are grouped in sections by diseases characterized by disturbances of metabolism of carbohydrates, amino acids, lipids, steroids, purines, metals, porphyrins and hemes, and connective muscle and skeletal tissues, haematopoietic system, epithelial transport and circulatory enzymes and plasma proteins.

It is the reviewer's impression that the presentation of genetic information and evidence has been improved in many chapters and the initial presentation of genetic theory has been expanded to three chapters; one of these by M. B. Hoaglund on information theory and protein synthesis is very helpful, particularly because, as the authors point out in their introduction, in many chapters it has been possible to attempt a molecular biological explanation of the disorders being considered.

It is remarkable that, in a book with so many chapters by different contributors, such a useful uniformity of presentation has been maintained without apparently inhibiting the characteristic styles of writing by the authors. Thus, most chapters start with a history of the discovery and development of knowledge of disorders to be described, and all end with numbered summary paragraphs which will prove most helpful as an orientation to those who are using the book as a source of reference.

The book will need no recommendation to those familiar with its predecessor. It is impossible to review the contents in detail even if any one reviewer was competent to undertake such a vast task. All that can usefully be said is that this is a unique book which has no rival in its scope and excellence, either as an introduction to many aspects of metabolism or as a source of reference to those working in most disciplines of biology and medicine.


Both these books are important—the first as a lucid and up-to-date textbook of genetic principles as applied to medicine, and the second as a thoughtful essay on the social implications of medical genetics. Both works call for close study.

The Thompsons of Toronto cover an exceptionally wide field in their 15 chapters, which are all concerned with basic considerations. Only the one chapter which deals with modes of inheritance follows the pattern of the older books. For the rest, there are discussions on the chromosomes and their aberrations, and on the molecular, biochemical, immunity, mathematical, and population aspects of human genetics. Other chapters deal with genes in differentiation and development, with studies on twins and with dermatoglyphs. The book is intended for the medical student and is offered as a contribution to the unsolved problem as to the place of genetics in the medical curriculum. It should find a ready welcome, from the post-graduate student and the inquiring clinician, but its catholicity is also likely to fire the enthusiasm of an occasional exceptional student.

Dr Smith's much shorter contribution also aims mainly at the undergraduate. It starts with the recognition of the facts that simply inherited characters are
unlikely to be common if they are seriously disadvantagous and that those genetic defects that are both common and severe are unlikely to be determined by simple hereditary mechanisms. For these reasons medical genetics is as yet of limited interest to students and to most clinicians. Though some elementary principles are expounded, the text is mostly devoted to the demonstration that medical genetics is making a major contribution to medicine by its recognition that genetic factors in disease are not only widespread but often brought out or masked by environmental factors, and that in consequence the management of disease is likely increasingly to depend on adjustment of individuals identifiable as being subject to special (genetically determined) risk. The manipulation of environmental factors in the control of genetic disease is of course no radical break with medical practice.


The first of these symposia was held at Cornell University in March 1965 to commemorate the centennial year of the founding of the University and the publication of Mendel's paper. The papers aimed less at giving new information than at indicating trends and new developments. The subjects covered were: the study of genetics in man—retrospect and prospect, by L. C. Dunn; certain embryological considerations, by Alberto Monroy; the future of mammalian cell genetics, by Robert S. Krooth; genetics and evolution in three human serum proteins, by Alexander G. Bearn and W. Carey Parker; the chromosomes, by Klaus Patau; and the application of knowledge, by John H. Edwards. The contribution by Krooth, which is exceptionally wide and adequate, occupies almost half of this issue.


If not a tract for the times, this book is a welcome signpost of trends in medical genetics. The authors of the Congenital malformations of the hand and their surgical treatment are orthopaedic surgeons who recognize that the concentration of attention to traumatic lesions of the hand no longer reflects current problems, and that the congenital anomalies present specific surgical requirements. The first half of the book is devoted to an excellent introductory chapter on malformations in general and to their experimental production, and this is followed by a competent survey of the commoner congenital and hereditary anomalies of the fingers and hand. There is also a useful tabulation of the syndromes in which hand defects occur. In all this no new ground is broken, but the second half of the book with its stress on the poor viability of malformed tissue and the need for minimal surgical trauma in operations is a thoughtful contribution, of value to the geneticist and still more to the surgeon to whom it is directed.


Since Apert in 1906 and Crouzon in 1912 described the syndromes that now carry their names, a voluminous literature on multiple anomalies has developed which has drawn not only on fresh observations but also on records in the older literature. Many of these observations show a consistent pattern, but a number fit ill into any pattern. While overlaps and formes frustes do occur, the classification of these defects presents considerable difficulties both clinically and pathologically. These difficulties are not helped by a mass of eponymous designations of doubtful validity. The greater part of the monograph by Tridon and Thiriet is devoted to the clinical aspects of the field covered by their review, and presents a welcome clarification of a confused literature. The opening section deals with the cranio-facial anomalies. These are discussed under four major headings: anomalies of the cranium, the cranio-facial skeleton, the facial skeleton, and cranio-cleido dysostosis. The authors rightly regard many of the anomalous cases as variants of a relatively few well-established entities and so help to bring some order in an apparently disorderly mass. A second section deals with the eye: this is particularly lucid, the literature being discussed under five headings: anophthalmos, coloboma, retinal anomalies, cataract, and neuromuscular defects. A concluding section deals with encephalopathies and is mainly devoted to Down's syndrome. The clinical part of the monograph is supported by some 350 references and these, though far from complete, are adequate.

In addition to these discussions, there are chapters on the embryology of the extremities and on experimental and clinical teratology. The genetic implications are indicated throughout the text. The over-all conclusion that there is no embryological basis for the association of anomalies of hands and feet with those of the head does not of course break new ground.