**Book Reviews**


As morbidity and mortality in childhood due to genetic causes increase, so the physician is increasingly asked for genetic advice. In disorders where the genetics are straightforward such advice might be given with advantage by the general practitioner or the paediatrician. However, with more complicated problems the parents are best referred to a genetic counselling clinic. In the former situation the adviser needs to be familiar with basic genetic principles and aware of the pitfalls in giving genetic counselling. This book was written to provide information which the family doctor might need to help parents who seek genetic advice. It was not intended for the specialist.

The material covered includes the 'tools for counselling' (drawing the pedigree, dermatoglyphs, cytogenetic studies etc.), modes of inheritance, including a brief discussion of multifactorial inheritance but with a separate chapter on X-linked inheritance, and counselling in chromosome abnormalities. Other chapters deal with twins, inborn errors of metabolism, the genetics of cancer, and mental retardation.

The chapter on inborn errors of metabolism includes a useful table in which is summarized information on the biochemical defects, incidence, symptoms, and therapy in some 29 disorders.

Throughout the approach is discursive, and the reader looking for facts will often be frustrated if not disappointed, and in some instances he will actually be misled. For example, the risk of a D/D translocation carrier having an affected child is not 33.4%, but considerably less. Other points of criticism are that marriage between cousins in general is discouraged, the importance of recognizing genetic heterogeneity is mentioned briefly and then only in regard to achondroplasia and diastrophic dwarfism, and there are errors of definition. For example, a syndrome does not represent manifestations of a 'defective complex of genes' because such a complex would have been disrupted by genetic recombination.

The book suffers from the serious drawback of not being sufficiently factual and would have been much improved if fact rather than opinion had been emphasized.

**Alan E. H. Emery**


Many severely subnormal children have to a greater or lesser degree abnormal facial features. Some of these, such as hydrocephalus or mongolism, are easily identifiable, but there are many rarer but important syndromes which may be diagnosed by inspection if the characteristic facies is recognized. This atlas presents, at one distressing glance, coloured photographs of the main features of some 80 mental retardation syndromes arranged in alphabetical order for easy reference. On the page facing the photographs in each case all the main physical signs, and the laboratory, x-ray, and other findings are summarized. The genetics of each condition, as far as is at present known, is given with appropriate references. There is a useful index of clinical characteristics which is detailed and runs to 18 pages. This book is well produced and should be consulted by those working in diagnostic clinics and residential institutions. Many of the syndromes described are often missed because of their unfamiliarity. Improved diagnosis and reporting of these conditions will assist those undertaking research into the genetic and pathological aspects of mental retardation. The old concept of mental deficiency as an entity is increasingly giving way to the realization that mental retardation is but a common symptom of many different specific physical abnormalities which need much more detailed study.

**L. T. Hilliard**


This expensive book is the product of a conference held already over a year ago. Many of the topics discussed were then, and still are, based largely on hypotheses which are subject to the impact of new experimental data. The book, then, can be regarded as only a landmark in a changing scene.

Well over one-third (245 pages) is devoted to brain circuitry and its structural basis and, apart from allusions to molecular genetics, is unlikely to be of central interest to those whose main concern is medical genetics. There is, however, a particularly interesting contribution by Paul Mandel on problems of genetic expression in nervous tissue; he discusses the fundamental topic of RNA synthesis and brain function. This theme is later developed by Holger Hyden in a section of brain research in learning and memory. The genetic potentialities of the brain are considered in terms of neuronal RNA and its effect on learning and memory. This topic has drawn an increasing band of investigators and the idea is attractive that a substance can serve as a memory mechanism both for innate behaviour and for experimental learning. Experimental evidence is discussed supporting the belief that in response to the establishment of new behaviour patterns there is synthesis within the brain cells of RNA of a highly specific base composition and of several types of acid proteins. The doubts that exist, however, concerning the unequivocal interpretation of such experimental results, are reflected in the discussion that follows this contribution.
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The next part, 'On the basis of disorder in the nervous system', begins with a sketchy section on inborn errors of metabolism. Much hope has been invested in the study of this group of conditions, many of which are identified specific enzymopathies, as a likely area in which to make discoveries concerning the interaction between biochemical change and psychological function. It is disappointing, therefore, that though the individual contributions are of high quality, they are pitifully few in number, and do not serve to cover this important field at all adequately within the context of a book devoted to the future of the brain sciences. James Austin, on inborn errors of glycolipid metabolism, provides particularly interesting matter regarding the direct bearing of altered metabolism on the structure and function of nervous tissue. This is getting close to the nub of the crucial problem of how genetically-determined biochemical disorders affect intellectual function, and it would have been fitting if more space could have been given to this theme.

A more appropriate allotment of space is given to the genetics and biochemistry of the psychoses. This opens with a full and critical discussion by James Stabenau of twin and family studies in schizophrenic and affective psychoses. This is followed by a number of competent reviews of biochemical changes that may be of aetiological significance in schizophrenia, involving mainly dimethoxyphenetyleamine, urinary indoles, and immunoglobulin. Similarly, changes involving the biogenic amines, electrolyte balance, and hormones in affective psychoses are discussed. These sections give a good survey of the areas in which leads are being sought to an understanding of the constitutional bases of the psychoses.

Like the proceedings of most conferences, this book suffers from a certain patchiness in emphasis on detail and lack of comprehensiveness, though the contributions are consistently of high quality. The diversity of disciplines represented also leads to a rather lumpy whole. It is a book, parts of which would be interesting to readers of very many different disciplines, but it is unlikely to give sustained interest to the medical geneticist unless he has a particular interest in one of the specialized lines of biological approach to brain research represented here.

Valerie Cowie


Most publications dealing with human chromosomal aberrations concentrate on the physical abnormalities. There is, therefore, added interest for a volume which attempts to assess the effects of abnormal chromosome constitutions on the minds of the patients.

The author, who is chief of the children's psychiatric service in the canton of Berne, set out to answer the question how far chromosomal aberrations correspond to specific psychiatric syndromes. The study is based on his own patients as well as a mass of published work which, with over 1200 entries, takes up a third of the pages. Even so, some important references are omitted. On the credit side, the text is very readable.

The author concludes that the psychiatric abnormalities produced by different chromosomal aberrations are non-specific. The results of different autosomal aberrations are such as would be found in any early-occurring brain damage. Moreover, if large numbers of sex chromosomes are present in excess, the effects produced are similar to those found in autosomal abnormalities.

The larger part of the text is concerned with the mental problems of patients with Klinefelter's and Turner's syndromes and their reactions to hormone therapy. The author thinks that the psychological differences between Klinefelter and Turner patients are largely due to their respective environments, because they will be judged by different criteria and, therefore, may be expected to react differently. Even if geneticists may not agree with all of Dr. Züblin's conclusions, they would do well to consider them.

Ursula Mittwoch


The choice of subjects and the high level of the contributions make this volume of Proceedings of exceptional interest. The complexities of the clinical aspects of muscular dystrophy were discussed by the President (J. D. Allan) and J. N. Walton, while the biochemical aspects were dealt with by R. J. Pennington, V. Dubowitz, B. McArdle, and P. Hudson. These papers and the discussions on them—excellently summarized—occupy half this volume and summarize admirably current knowledge and the problems at issue. The rest of the text roams widely over strictly biochemical studies (such as those by Raine and by Jatzkewitz on sphingolipidoses) and on biochemical lesions in such affections as Wilson's disease (J. M. Walshe) and inherited brain disorders (E. M. Brett). Well edited and well produced, this volume is more than a report of transient interest.

Arnold Sorsby


This book is the product of an International Symposium on Diagnosis and Treatment of Disorders Affecting the Intrauterine Patient, held at Dorado, Puerto Rico, 29–31 October, 1967, in commemoration of the bicentennial of the Department of Obstetrics and Gynecology, College of Physicians and Surgeons,