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 patients 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%, 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 acidic proteins. The doubts that exist, however, concerning the unequivocal interpretation of such experimental results, are reflected in the discussion that follows this contribution.
Atlas of Mental Retardation Syndromes. Visual Diagnosis of Facies and Physical Findings
L. T. Hilliard

J Med Genet 1970 7: 102
doi: 10.1136/jmg.7.1.102-a

Updated information and services can be found at:
http://jmg.bmj.com/content/7/1/102.2.citation

These include:

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/