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BOOK REVIEWS

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Human Malformations and Related Anomalies. Volumes I and II. Oxford Monographs on Medical Genetics No 27. Editors R E Stevenson, J G Hall, R M Goodman. (Pp 265, 1162.) Oxford: Oxford University Press. 1993.

The real measure of a text book is whether it is used, so I decided to delay this review a little to see how often I consulted it in the course of my clinical work. My main excuse for a somewhat longer delay, however, is that the book was borrowed so frequently by my colleagues that I was constantly chasing it to consult and to review. I am pleased to say that these "field trials" have confirmed without doubt the excellence of this huge and scholarly work. It will be a key reference text for students, researchers, biomedical scientists, and clinicians, particularly clinical geneticists, paediatricians, and obstetricians.

Volume I is the smaller (265 pp) and comprises 15 chapters, mainly overviews, which fall into two main categories. The first concerns introductory clinical and basic science topics, for example, terminology, embryology, epidemiology, and the genetic and environmental basis of human anomalies. The second category deals with non-technical aspects of human malformations such as historical perspectives, cultural and societal dimensions, and counselling.

Volume II is largely organ or systems based, though there are chapters on twins, asymmetry, and pigmentary disorders. Most chapters begin with a brief embryological and anatomical description and then various malformations are defined, described, and illustrated in detail. For almost all malformations, syndromes which show the particular anomaly are tabulated. Each entry ends with aspects of treatment, prognosis, and prevention. Extensive and up to date references are given and the vast number of illustrations and line diagrams are excellent.

The standard of presentation and content of all chapters is good but two particularly excellent entries will be described to persuade potential purchasers of the good sense of an investment. The chapter on the brain is divided into 14 sections, each describing a particular type of abnormality such as microcephaly, megalencephaly, holoprosencephaly, neuronal formation, and migration disorders. Tables of syndromes with the particular anomaly are included and key features and references given. Illustrative material used includes clinical photographs, macroscopic and microscopic pathological sections, CT, MRI and ultrasound scans, x rays, line diagrams, and flow charts. The chapter on the ventral wall of the trunk begins with an extensive review of embryology highlighting timing and location of errors leading to malformations. This approach means that what could have been a descriptive "rag bag" of anomalies hangs together in a most interesting chapter which describes among others, sternal defects, umbilical anomalies, abdominal wall defects, bladder exstrophy, and breast anomalies.

The editors are to be congratulated on their concept and design of these volumes. Enormous care has been taken to ensure a uniformity of approach which, however, does not detract from the individual style of the various contributors. It is customary for reviewers to identify errors or weak parts of books but I really don't have any criticisms. Perhaps the authors could have speculated more on the molecular basis of malformations, or referred more often to homology with malformations in the mouse, but then again perhaps this book provides just the right amount of information to stimulate the readers themselves to think and look further. I would strongly recommend that this book is used as a "bench book" and is located in all clinical genetics departments; the hospital or university library should also contain a reference copy.

DIAN DONNAI

Genetic Studies in Affective Disorder: Overview of Basic Methods, Current Directions and Critical Research Issues. Editors Demitri F Papolos, Herbert M Lachman. (Pp 236; £32.95.) UK: Wiley-Interscience Publications. 1994.

The likely advances in determining the aetiology of major mental illness that are predicted for the next decade will receive a substantial contribution from the application of molecular biology to genetic studies. Consequently there is a pressing need for researchers in the field to communicate their subject in a way that is accessible and relevant to those in the front line of mental health care delivery. This book is an attempt to do so and it succeeds, although to varying degrees.

This is the eighth title in the Einstein Psychiatry Series of the Albert Einstein College of Medicine, the aim of the series being to record developments and achievements in psychiatry and, where appropriate, to present disparate viewpoints within the profession. The targeted readership is psychiatrists, psychologists, social workers, and health care workers in general, an ambitious cross section for what is, at times, a highly technical and focused text. The list of contributors is impressive and contains many of the leading names in genetic studies.

The book is divided into three sections; part one covers basic methods and critical research issues in linkage studies and, for me, is the primary strength of the book. The chapters on genetic epidemiology and diagnostic issues in pedigree assessment are excellent, being comprehensive yet concise, and attesting to the editors' claim of little previous genetic education being necessary. I suspect that this is not wholly true for the subsequent account of basic principles in linkage analysis. It is a lucid rendering of molecular biological methods – I particularly appreciated the correlation of techniques with laboratory man hours – but the truly uninitiated may need a more basic genetics textbook for cross reference. Janice Egeland's chapter on her pioneering study among the Old Order Amish gives a fascinating glimpse of the story behind the headlines and is a sobering reminder of the many pitfalls that may be encountered by the psychiatric geneticist.

Part One concludes with a summary of the findings of molecular genetic studies to date, except that the date appears to be no later than 1991 and, given the remarkable speed at which genetic studies are progressing, a three year delay between writing and publishing must now be considered unacceptable. Julian Mendlewicz makes claim for an X linked form of bipolar illness; likely it may be, but further studies have failed to replicate linkage with DNA markers in new pedigrees and the initial excitement generated at the time of his writing is now somewhat tempered by a number of unresolved issues.

The middle section is devoted to clinical aspects of genetic studies. Working from the premise that familial illnesses need a familial model of treatment a proposed psychoeducational approach is outlined and over four pages are designated for clinical examples. This merges into the next chapter on genetic counselling issues which uses the orthodox Jewish community as an example throughout. There is a remarkable degree of repetition within the two chapters (which are the longest in the book) and, although there is some interesting debate around psychological and ethical issues raised by counselling, the frustrating reality is that the facts generated in Part One are too scant to allow useful application in Part Two: at present we're still asking the questions!

Part Three looks to future directions in research, focusing on two specific areas: animal models and in vitro systems. The former consists of a report of a proposed animal model of depressive illness (so called "learned helplessness") derived from Beck's cognitive theory of depression, and shows how these models can help to tease apart the effects of environmental stressors and genetic vulnerability. The latter uses the example of Lithium's mode of action to show how gene expression may be modulated at a molecular level.

The reader would require some understanding of biochemistry to follow the involved, albeit persuasive, argument. A more general overview of proposed mechanisms controlling gene expression may have been more accessible and enlightening to a broad readership and would have allowed some discussion of pertinent topical issues such as anticipation and genomic imprinting.

In summary, this is primarily a book for psychiatrists new to molecular genetics. It will provide them with a useful introduction to principles and techniques but they must be aware that in terms of research findings it is already out of date. It amply indicates the particular difficulties of genetic studies in psychiatric disorders but, understandably, assumes a working knowledge of mental health pathology that may limit its usefulness to non-psychiatric geneticists.

STEPHANIE SADLER