BOOK REVIEWS

Catalog of Teratogenic Agents

10th edition. Thomas H Shepard. Baltimore: The Johns Hopkins University Press. 2001. ISBN 0-8018-6722-3.

This book is a cornucopia of information about potential or proven teratogenic effects of drugs and other agents. It is not only a delight for aficionados of catalogues, but also an essential addition to the library of all clinicians who deal with would be or pregnant mothers, management of illness in pregnancy, and the management of children with congenital disorders. Although the major emphasis is on pharmaceutical agents, some herbal remedies are also included, as are recreational drugs, viral infections, maternal illnesses, environmental chemicals, radiation and radioactive chemicals, and some genes. The book sets out to answer the question, "Does this agent produce congenital defects in the human or animal", and goes a long way towards fulfilling its goal within the limitations of present knowledge. Of course, there are gaps in the catalogue; for example, some of the newer antiepileptic drugs do not appear (topiramate, tiagabine), but then there is little or no published information about their use in pregnancy. In some cases, perhaps even an entry stating that there is no information might be useful. As the mission statement suggests, the book concentrates on malformations, so there is less than one might hope on neurodevelopmental disorders following exposure to some drugs. UK readers should also be aware that drug names are taken from the Merck Index, and these do not always correspond with the names in common usage through the British National Formulary. However, for those which I checked, the British name was cross referenced through the index. The catalogue comprises some 3073 entries, including useful sections reviewing evidence for the teratogenic effects of vitamin deficiencies. The folic acid section is quite extensive, and makes reference to recent data concerning congenital heart disease, urinary tract abnormality, and orofacial clefting as well as the better known association with neural tube defects. Some gene disorders are also included, such as *Hox* genes and *FGFR3*, but curiously there is no specific section dealing with genes encoding drug metabolism enzymes, such as the cytochrome P450 enzymes. The methylene tetrahydrofolate reductase gene (mutation in which has been associated with risk of neural tube defect) appears in the folic acid section, but has no separate section of its own and is not referenced in the index. Despite these idiosyncrasies in the catalogue, the author is to be congratulated on producing such a near comprehensive work, and the book also includes some useful short introductory chapters on definitions in teratology and a comparative table of timing of embryological

events in humans and experimental animals. It is very much an evidence based work, with an extensive bibliography of original publications. All in all, this is an essential reference book for clinical genetics, as well as for other specialties. With the rapid changes in drug therapies and the information explosion affecting teratology as well as other branches of genetic medicine, an electronic version with improved indexing, or an online version would be welcome. Although this is apparently available as part of a larger software package, technophiles may be disappointed that this catalogue is only available by itself on paper.

John C S Dean

Emery and Rimoin's Principles and Practice of Medical Genetics

4th edition. Editors D L Rimoin, J M Connor, R E Pyeritz, B R Korf. (Pp 4936; £350.00.) Edinburgh: Churchill-Livingstone. 2002. ISBN 0443064342.

It is almost 20 years since the first edition of this book became available and, in that time, there can hardly be a department of clinical genetics anywhere that has not felt the need to acquire a copy. It has carved a special and much respected place in our professional affections. This new edition, after a gap of over five years of momentous scientific discovery and relentless reshaping of clinical practice, will require the provision of very significant shelf space, stretching, as it does, to three packed volumes and offering not just an updating of previous work, but the addition of over 20 new chapters.

They are virtually all represented within these covers - the great and the good among geneticists, some famous, others awaiting the call of fame, but common to all contributors is a track record of distinction within their field of expertise. The general layout of the book is broadly similar to that of its immediate predecessor, with an initial 21 chapter section on "Basic Principles", followed by a new section entitled "General Principles," comprising 13 chapters. Thereafter, follow nine chapters comprising the section entitled "Approaches to Clinical Problems." The last section, "Approaches to Specific Disorders" accounts for over 100 chapters, each considering groups of related conditions, such as "cardiomyopathies" or "renal cystic diseases".

In contrast to reviews of most other books, one cannot hope to read the book from cover to cover, check occasional facts, smile or scowl as appropriate, and reflect the experience in 250 words. I have read individual chapters, randomly chosen either for the interest of the title ("Medicine in a Genetic Context" in which Dr Barton Childs offers interesting ideas not often presented to trainees and worthy of every clinician's attention), or for personal clinical and research interest ("Craniosynostosis" in which Drs Lewanda and Jabs do well to summarise a difficult and potentially confusing body of publications into a sensible overview, while betraying a highly selective and unrepresentative approach to citation of relevant published reports), or simply to address my absolute ignorance ("Twins and twinning", in which Dr Judy Hall stimulates, excites, and informs in equal measure). Additionally, these volumes have rested on the window sill next to my desk for several weeks allowing

me ready access in those moments of blankness that are the daily lot of the jobbing geneticist. It has been my experience that there has usually been some clue or idea to be found there that enabled one to address the clinical situation with a structure and purpose that might otherwise have been lacking. This observation is especially true in relation to the investigation of particular clinical presentations. It has been no less valuable at reminding myself of those essential facts, once clear, but which have become dimmed with time and the grasp of which is now recognisably tenuous.

It has to be asked whether, in an age of highly specialised databases, subspecialist textbooks, and widespread online access to multiple sources of information including clinical guidelines, a book of essentially general nature still commands a place. It is true that much of the information offered herein is of general nature and subspecialist reading in the journals or other sources will often be required in respect of individual patients. Nonetheless, to have brought together between the covers of three volumes a comprehensive summary of current thoughts and practices in clinical genetics as practised in the leading departments across the world by some of the greatest geneticists is a magnificent achievement. It is a body of experience from which we can all learn and to which I suspect most of us will continue to turn on a regular basis. So, although this book will be supplemented by specialist resources in every department, there cannot be a medical library or a department of clinical genetics which can afford to be without it. It should form the backbone of many a medical genetics library. It is well finished in high quality paper, beautifully bound, as it needs to be, and a credit to editors, authors, and publishers alike.

Willie Reardon

Genetics for Cardiologists: The Molecular Genetic Basis of Cardiovascular Disorders

Ali J Marian. London: ReMedica Publishing. 2001. ISBN 1 901346 09 9.

Nowadays, you can hardly open a cardiology journal without finding at least a few papers describing the latest advances in one or other aspect of the genetics of cardiovascular disease. For those not familiar with the terminology or techniques, enjoyment and appreciation of the, often excellent, underlying science can be a frustrating experience. This short book of less than a 100 pages (including glossary), one of a series of monographs covering various clinical specialties, provides a digestible introduction to the field of genetics and a summary of the findings for both monogenic and complex cardiovascular disorders as things existed in 2000. A brief description of commonly used genetic analysis techniques is also given.

The familiar cardiovascular disorders with a well established genetic basis, such as hypertrophic cardiomyopathy, long QT syndrome, hypertension, and atherosclerosis, are all covered. However, many cardiologists will be surprised to learn, as I was, of the existence of familial forms of a myxoma syndrome as well as patent ductus arteriosus. In pursuit of uniformity, an attempt is made to discuss the data on each disorder using the same sub-headings (for example, mutational spectrum, effect of mutation, molecular pathogenesis,

genotype-phenotype correlation, etc.) This is only partly successful because for many of the conditions, information on one or more of these aspects remains unknown. A brief clinical description of each condition is given. For a cardiologist, this information is probably unnecessary. Although the book is aimed at this readership, I suspect that it will perhaps be most helpful to young scientists working in the field of genetics of cardiovascular disease who need a general introduction to the field, and especially the important clinical aspects.

However, there are a few errors, such as the statement that onset of familial atrial septal defect occurs in the third and fourth decades when it is, of course, congenital. A limitation is that references to original articles are not given.

I enjoyed reading the book and compilation of genetic data on all diseases related to a particular system has much to commend it, for both the specialist as well as a general reader. However, the genetic field is moving rapidly, nowhere more so than in cardiology. There-

fore, through no fault of the author, some of the data are already out of date and exciting new discoveries, for example, the finding that mutations in the bone morphogenetic protein receptor type II underlie some familial forms of primary pulmonary hypertension, are not covered. To be most valuable, this sort of book needs to be regularly updated. In this electronic age, this should be possible. Dr Marian has made a good start and I hope he feels motivated to produce further editions.

N J Samani

Want to extend your search?

Cross journal searching

If you can't find what you are looking for in the *Journal of Medical Genetics* you can extend your search across many of the more than 200 journals available for selection. You can restrict your search to specific subject areas (eg, clinical medicine, basic research), or select specific journals, or search all available titles.

www.jmedgenet.com