BOOK REVIEWS

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Nothing better illustrates the exponential rise of interest in birth defects and syndrome identification during the last decade than the almost simultaneous publication of these two enormous volumes. Together with the recently published third edition of Syndromes of the head and neck (reviewed in the December 1991 issue of the Journal) they provide a unique data source for the clinical geneticist/dysmorphologist by extending the range of possible textbook diagnoses far beyond that at present available in the standard syndrome atlases.

Birth defects encyclopedia is in all but name the third edition of the Birth defects compendium first issued in 1973. Based on the records of the Center for Birth Defects Information Service, it contains brief details of over 2000 congenital malformations and multiple malformation syndromes, along with a curious pot-pourri of seemingly inappropriate entries, such as rheumatoid arthritis and susceptibility to diphtheria. Each condition is discussed using over 20 standard headings, most notably clinical findings, complications, and aetiology, with due regard to mode of inheritance and gene mapping information where appropriate.

Compiling the data and contributions for this multi-author megatome must have been a mammoth task, so perhaps it is unreason- able to hint at criticism of such a major undertaking. Yet it is difficult not to focus on some of the more obvious shortcomings. Its sheer size and weight (4.8 kg), that is, well above the 97th centile even for genetic textbooks, make it very unwieldy, and the absence of an index is a particularly frustrating omission. Many of the entries are authoritative, well illustrated, and entirely apposite, but some are rather brief and incomplete.

The sections on recurrence risks are often totally superfluous given that most readers will be well acquainted with the basic principles of mendelian inheritance.

In contrast, Multiple congenital anomalies assumes a very well informed readership and makes no pretense to be anything other than an aid to diagnosis for dysmorphic syndromes. The information provided is drawn straight from the records of the London dysmorphology database which is itself now available commercially through the electronic division of Oxford University Press. Like the Birth defects encyclopedia, this book suffers from a surfeit of weight (3.95 kg), but in other respects it is much more difficult to criticise, this possibly reflecting its uniformity of authorship, unlike the Encyclopedia which draws on the experience of over 1000 contributors.

Essentially, Multiple congenital anomalies consists of two main sections, the first being a catalogue of approximately 2000 syndromes each described under the four headings of mode of inheritance, abstract, features, and references. There are no frills or illustrations, nor is there an index. The second section is by far the more valuable and takes the form of a diagnostic database. Inevitably this lacks the versatility of the full computerized program that will produce useful clinical diagnostic handles, a manageable selection of differential diagnoses can be drawn up quickly and easily without recourse to the microchip.

As the authors rightly state in the preface, computers are not always as user friendly or reliable as their manufacturers would have us believe, so that this book is certainly a useful addition to the busy clinical geneticist's diagnostic arsenal, particularly when faced with a telephone enquiry or abnormal infant on a special care baby unit. Access to the full computerised version of the database is a sine qua non, but for those with limited time or who are stranded in a satellite clinic this is a very worthwhile substitute.

Ideally every department of clinical genetics should have access to both of these books which complement rather than compete with one another. If I had to choose between one of these and Syndromes of the head and neck, then I would perhaps opt for Dr Gorlin's magnum opus, but it could reasonably be argued that comparisons are invidious and if funds are sufficient then all three of these superb books should grace the shelves—suitably reinforced—of every departmental library.

I D YOUNG


Five of this year's 24 reviews are about human genetics: two pairs of articles on mutation rates and on heart disease, and Eric Stanbridge's review of 'Human tumor suppressor genes'. The latter field moves so fast that it must be difficult to review, but Stanbridge provides a useful perspective by placing the recent cloning successes against the background of cell fusion experiments extending over many years.

On heart disease, Singh and Moll give the broad view: 'Genetics of atherosclerosis'. They contrast the "top-down" strategy of segregation analysis with the "bottom-up" strategy of linkage and association studies. I remain to be convinced that the top-down approach tells us anything useful about heterogeneous traits, but then it could be argued that the record of linkage and association studies is also too glori- ous. When atherosclerosis, which is adequately understood biochemically, is proving so hard to analyse, it is a brave person indeed who tries to do the same with unknown territory like the heart. This review is partnered by an excellent article from Hobb's, Russell, Brown, and Goldstein on 'The LDL receptor in familial hypercholesterolemia'. The molecular pathology is discussed in the light of the many characterised LDL receptor mutations, and the role of thu sequences in the generation of deletions is stressed. A very useful summary.

Perhaps the most important review in the book is by Neel and Lewis on 'Comparative radiation genetics of humans and mice'. The authors attempt to estimate the doubling dose, that is the amount of acute or chronic radiation that will produce a detectable biological effect in a population. This is in all likelihood a gross overestimate if one compares it to the acute dose. Perhaps the authors intended their figures as a guide to the important question of stochastic versus hereditary effects on populations, but the very large uncertainties in their estimates are not appropriate as a basis for a national policy on radiation exposure. The book is written in a very lucid style, and I can hardly criticise the authors for being wrong. One can only hope that future reviews will benefit from the authors' experience and that the field will not move so fast that it is impossible to keep up.

As always, much of the pleasure of Annual Reviews comes from dipping into articles on areas of more peripheral interest to the clinical geneticist. I would particularly mention Kingsbury on 'Genetics of response to slow virus (prion) infection', Katriny Shaiya on 'Generation of diversity in retroviruses', and Atwater et al on 'Regulated mRNA stability'. You can rely on Annual Reviews being well written and authoritative, and the whole package comes at a very reasonable price, especially for members of the American Society of Human Genetics. Definitely worth getting.

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