

BOOK REVIEWS

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Birth Defects Encyclopedia. Ed M L Buyse. (Pp 1892; £150.) Dover, MA: Center for Birth Defects Information Services/Oxford: Marston Book Services. 1990.

Multiple Congenital Anomalies. R M Winter, M Baraitser. (Pp 1433; £175.) London: Chapman and Hall. 1991.

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 unreasonable 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 pretence 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 computerised program, but with careful choice of 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 probably 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

Annual Review of Genetics. Vol 24. Ed Allan Campbell. (Pp 714; \$38.00.) California: Annual Reviews Inc. 1990.

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, Sing and Moll give the broad view with an article on '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 not too glorious. 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 psychoses. This review is partnered by an excellent article from Hobbs, 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 Alu 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 the same mutational impact on a population as occurs spontaneously each generation. Their human data come entirely from the studies in Hiroshima and Nagasaki. As is well known, there is so little evidence of mutations among children of the survivors that even huge studies conducted over many decades provide only the vaguest estimate of the doubling dose. Neel and Lewis present a great deal of data, set out the arguments and problems extremely clearly, and conclude that the chronic doubling dose for both humans and mice is around 4 Gy. This, of course, leaves the Sellafield and Dounreay leukaemia clusters totally unexplained. This review should be required reading for anyone who believes that 'the facts' are simple.

Related to this is an interesting review by Albertini and colleagues from Vermont on 'In vivo somatic mutation in man'. This discusses the mutation rates estimated by screening large numbers of red cells or T lymphocytes from a few subjects for the occasional cell showing the sickle mutation, or mutations in the MN blood groups, HPRT, or HLA products. These systems typically show five to 30 mutations per million cells in adults. Interindividual variations exist, and might be relevant to susceptibility to somatic genetic diseases such as cancer. Mitotic recombination accounts for 30% of HLA mutations, but interestingly there is no evidence for gene conversion, even though this is a principal mechanism of mutation in the 21-hydroxylase genes next door.

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', Katz and Skalka 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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