Journal of

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BOOK REVIEWS

All titles reviewed here are available from the BMJ Bookshop, PO Box 295, London WC1H 9TE. Prices include postage in the UK and for members of the British Forces Overseas, but overseas customers should add 15% to the value of the order for postage and packing. Payment can be made by cheque in sterling drawn on a UK bank, or by credit card (Mastercard, Visa, or American Express) stating card number, expiry date, and full name.

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.

ANDREW P READ

Notice to contributors (general guidance)

The readership of *Journal of Medical Genetics* is world wide and covers a broad range of workers, including clinical geneticists, scientists in the different fields of medical genetics, clinicians in other specialities, and basic research workers in a variety of disciplines. It publishes original research on all areas of medical genetics, along with reviews, annotations, and editorials on important and topical subjects. It also acts as a forum for discussion, debate, and information exchange through its Letters to the Editor columns, conference reports, and notices. The editor is always grateful for suggestions or criticisms from readers and authors.

ORIGINAL PAPERS

These may be on any aspect of medical and human genetics and may involve clinical or laboratory based and theoretical genetic studies. Guidance on length can be obtained from studying the Journal. Shorter articles may be most appropriately submitted as *case or family reports*, not exceeding 1000 words, with no more than three figures, one table, and 10 references. *Short reports* should not exceed 500 words, with a single illustration. Contributions may also be submitted as *Hypotheses*, *Technical Reports*, *or Short Communications*. Accelerated publication of papers of particular importance will be considered.

REVIEWS

Short or longer reviews on all aspects of medical genetics are welcome, but should be discussed first with the Reviews Editor. Contributions on historical topics, or which could form part of specific series, are particularly acceptable.

ANNOTATIONS AND EDITORIALS

These are written or commissioned by the editors, but suggestions are welcome regarding possible topics and authors.

LETTERS

These are welcome on any relevant topic and will be published rapidly. Those relating to or responding to previously published items in the Journal will be shown to those authors, where appropriate. Although a paper submitted as an original report may sometimes be published in shortened form as a letter, it is preferable for initial submissions to be as a short report, unless directly related to a previous journal article.

CONFERENCE REPORTS

Reports from small to medium sized meetings, especially international workshops on specific topics, will be appreciated. Authors intending to submit conference reports should liaise with the Reviews Editor to avoid duplication.

SPECIAL ISSUES AND SUPPLEMENTS

These are published at intervals on topics of particular relevance. Enquiries are welcome from those organising workshops or symposia who may have material suitable for such an issue.

BOOK REVIEWS

The Journal aims to review as wide a range of relevant books as possible. Authors or others wishing to check if a book has been received may check with the Journal office. Computer programs and databases, official reports, and other material relevant to the field may all be appropriate for review. Enquiries about such items are welcome.

OBITUARIES

The Journal would like to be informed rapidly of the death of any senior or important person in the field of medical or human genetics, regardless of geographical location. In general, a brief notice would be published rapidly, with a longer obituary as appropriate. Since such deaths often occur many years after retirement, it will be appreciated if readers will contact the Reviews Editor so that appropriate arrangements can be made.

NOTICES

Notice of forthcoming meetings in different countries should be sent as far ahead as possible. Extensive descriptions should be placed as advertisements.

'CALLS FOR PATIENTS'

The Journal receives an increasing number of requests to publish notices of proposed studies involving patients or families with rare genetic disorders. In general such notices are appropriate only for major international collaborations; the proposer should ensure that such a notice does not conflict with existing studies or proposals.

ILLUSTRATIONS

High quality black and white photographs are preferred for most illustrations, particularly of patients. Colour illustrations can be accepted; however, authors are asked to pay part of the cost, so their desirability should be discussed in advance of submission. All identifiable photographs of patients must be accompanied by written permission for use.

Specific instructions to authors

Papers, which should be in triplicate and in the Vancouver style (*BMJ* 1988;**296**:401–5), should be sent to the Editor, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR and not to individual editors, with the exception of papers from the USA, which can be submitted to the North American Editor, Dr P M Conneally, Department of Medical Genetics, James Whitcomb Riley Hospital for Children RR129, Indiana University Medical Center, Indianapolis, Indiana 46223, USA. Submission of a paper will be held to imply that it contains original work which has not been previously published. It is the responsibility of the submitting author to ensure that all co-authors are agreeable for their names to appear on the manuscript. A FAX number should be provided. Permission to republish must be obtained from the Editor.

Where a patient(s) with a structural chromosome abnormality is described, the availability of a cell line(s) should be stated in the text together with its identifying number, cell bank, and, where appropriate, contact person.

All contributions should be accompanied by an abstract (preferably structured) giving the main results and conclusions. Typescripts should be at least double spaced with wide margins. One page proof will be sent to the author submitting the paper and alterations on the proof, apart from printer's errors, are not permitted. Reprints may be ordered when the proof is returned.

Figures should be kept to a minimum and should be numbered consecutively in Arabic numerals. Legends should be typed on a separate sheet.

Tables should not be included in the body of the text, but should be typed on separate pages and numbered with Arabic numerals. A legend should be provided.

References should conform precisely to the style current in this journal. Authors are responsible for the accuracy and completeness of their references as these will not be checked by the Editorial office.

NOTES ON NOMENCLATURE

Authors should refer to the following publications.

(1) Chromosomes: *ISCN 1985. An international system for human cytogenetic nomenclature*. Basel: Karger, 1985.

(2) Genes: Shows TB, *et al.* In: Human Gene Mapping 5 and 7. *Cytogenet Cell Genet* 1979;**25**:96–116, 1984;**37**:340–3.

(3) Loci: Conventional nomenclature should be used, with lower case lettering as appropriate (for example, Race RR, Sanger R. *Blood groups in man. 6th ed. Oxford, London: Blackwell, 1975; and Giblett ER. Genetic markers in human blood.* Oxford, London: Blackwell, 1969).

(4) Blood coagulation: International Committee of Haemostasis and Thrombosis (Graham JB, *et al.*). A genetic nomenclature for human blood coagulation. *Thromb Haemostas* 1973;**30**:2–11.

(5) Enzymes: *Enzyme nomenclature: recommendations of the nomenclature committee of the International Union of Biochemistry*. New York: Academic Press, 1984.