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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.

Human Gene Mapping 11. The Eleventh International Workshop on Human Gene Mapping. Ed Ellen Solomon, Chris Rawlings. (Pp VIII + 2200; £259.60.) Basel: Karger. 1992. (*Cytogenet Cell Genet* 1991;58:Nos 1–4, 1–2200.)

This report, contained in two volumes and amounting to 2200 pages in all, represents both a landmark and perhaps the end of an era. Since the report of the first human gene mapping workshop held in 1973, successive volumes have faithfully charted the development of the emerging human gene map, from the fragmentary data based on protein markers of the 1970s, through the explosion of new information from DNA markers in the 1980s, to the present situation where every chromosome has a map that is at least an outline, while in some cases it is already detailed.

The 1991 London meeting is likely to have been the last in the traditional form, where the heroic efforts of chromosome committee chairmen and behind the scenes computer staff allowed a combination of data entry, synthesis, and presentation and resulted in a unique flavour which was certainly exciting to those directly involved, though perhaps unnecessarily frenetic to those attending mainly as observers. The detailed and usually dry format of the reports has hidden (wisely) the emotion and exhaustion engendered by computer failures (remarkably lacking in the London workshop), disputes as to whether assignments were proven or provisional, and (inevitably) the question of appropriate nomenclature. While some of these issues will continue to arise in future individual chromosome workshops, the international camaraderie (and rivalries) and the general sense of a map painfully emerging before one's eyes will be missed by all dedicated gene mappers.

The first of the two volumes in this report begins with a catalogue of mapped genes which essentially represents the overall summary of our current knowledge on actual genes, as opposed to anonymous DNA segments. At 2325 genes, with the list covering 90 pages, this shows what a considerable way we have come, even though this total represents only around 5% of the estimated 50 000 human genes. The rest of volume 1 is entirely occupied by the report of the individual chromosome committees, with brief text reports indicating new information and tables giving the details of known genes and markers on each chromosome, representing an essential source of information for workers on any specific chromosome. Looking to the future it is this part that will largely become replaced by on line and continually updated databases.

Volume 2 contains the chapters from the committees dealing with clinical disorders, chromosomes and neoplasia, comparative mapping, and mitochondrial DNA, among others; it also has the abstracts of posters shown at the meeting. It is this volume that will be most valuable to clinicians and scientists in genetics overall, as opposed to gene mappers, though much space is also occupied by lists of anonymous DNA segments. The clinical report has relied extensively on the French Genatlas, which actually presents information in a way much more suited to workers on diseases than is the case with the rest of the volume.

A few points can be criticised in this report: from the viewpoint of the clinical geneticist it is supremely user unfriendly, despite the attempts of the clinical committee's report to overcome this – but then the workshops were never really aimed at those interested in genetic disease, even though clinically orientated workers formed an increasing proportion of those involved. Those who wonder by what miracle of production such a massive report on a meeting inAugust 1991 could be published by the end of the year (when only received for publication on 23 December) will be disappointed: it actually appeared in April 1992. Hopefully this will not presage a new policy of editors to try and improve their image of rapid publication and establish the priority of their authors by backdating all their books and journals!

Such criticisms should not detract from the value of this report, whose volumes will (and in the reviewer's case already are) much more often be found in use on someone's desk or bench than sitting on a shelf. If this is indeed a report on the last Human Gene Mapping workshop, then this is an occasion to salute not only the publishers and editors, but the whole of the devoted, disparate, and idiosyncratic community of gene mappers from many countries who have taken this field from its origins to its present flowering. Let us hope that, with commercial interests, robots, and politicians lurking in the wings to take over the human genome project, the spark of individuality which has characterised the field of gene mapping until now will be neither lost nor forgotten.

PETER S HARPER

The Cystic Kidney. Ed K D Gardner Jr, J Bernstein. (Pp 444; £95.00.) Dordrecht: Kluwer Academic Publishers. 1992.

Renal cysts, a common finding in clinical practice, occur in a multitude of inherited and acquired conditions. This multi-disciplinary book is an important contribution that will be a useful resource for those with an interest in renal cysts and the disorders with which they are associated. The editors have organised the text to carry the reader from an understanding of the structure and pathogenesis of renal cysts through to the diseases that are associated with renal cyst formation. They have also taken care with the selection of contributors, all of whom are well known from their previous contributions to renal cystic publications.

The clear structure of the book together with detailed, well referenced text makes it useful for a relatively wide audience. It has an obvious appeal to clinicians who will find great utility in this resource which provides numerous insights into the pathology, pathogenesis, genetics, diagnosis, and management of cystic kidney disease. The focus on management is particularly useful. High quality reproduction of photomicrographs and renal imaging further increase the utility of this book. Those involved in renal cystic research will also appreciate the perspective provided by this book in which the results of recent research have been well integrated into the wider clinical picture. This is particularly in evidence with the focus given to advances that have come from the application of human molecular genetics. However, fast moving developments in molecular biology mean that this section is quickly changing into an interesting piece of history.

While the numerous contributors have provided a useful depth of analysis, there is also an occasional conflict of opinion that is instructive to the reader. One small price for the benefits of having multiple contributors is a tendency for repetition. Overall, this collection of essays is an excellent summary of contemporary knowledge about renal cysts and the conditions in which they occur. For those with either a patient based or research based interest in renal cystic disease, this book is a rich resource that is strongly recommended.

DAVID RAVINE

Polycystic Kidney Disease. Contributions to Nephrology Vol 97. Ed M H Breuning, M Devoto, G Romeo. (Pp 142; £61.80.) Basel: Karger. 1992.

Funding from the Commission of the European Communities, Directorate Biology (COMAC-BIO) to generate close collaboration between research groups working on polycystic kidney disease was the stimulus behind this collection of review articles and research papers. Each paper was presented at the 2nd International Workshop of the European Concerted Action Towards Prevention of Renal Failure caused by Polycystic Kidney Disease, Parma, September 1991.

The stated aim of the workshop was to bring together researchers adopting diverse approaches towards polycystic kidney disease. As someone who did not attend the workshop, I found that these written proceedings do reflect this aim. The contributions include reviews of clinical interventions that have potential to slow down the deterioration of kidney function, a review of several experimental models of cystic kidney disease, including more recent transgenic and chimaeric mice models as well as animal models showing dominantly inherited cystic disease. The pathogenesis of cysts is reviewed, as is the current knowledge surrounding the molecular genetics of PKD1. The important issue of genetic heterogeneity is addressed, while some attention was focused on the advances in molecular and computing tools that will be of assistance in future studies on polycystic kidney disease.

Overall, this publication is a useful collection of papers that is an informative current resource for both clinicians and research workers with an interest in autosomal dominant polycystic kidney disease.

DAVID RAVINE

NOTICE

6th International Workshop on the Fragile X and X linked Mental Retardation

The meeting, to be held in Cairns, North Queensland, Australia, on 3–6 August 1993, will focus on all aspects of research into fragile X syndrome and other forms of X linked mental retardation. It is designed primarily for those actively involved in research in these areas and all participants will be expected to contribute actively to the workshop format of the meeting. For more information please contact: Dr Grant R Sutherland, Department of Cytogenetics and Molecular Genetics, Women's and Children's Hospital, North Adelaide, South Australia. Tel: (08) 204 7333 or fax: (08) 204 7342.

NOTICE

Medical Genetics: 1993

Board Review Course. On 21 to 23 May 1993, the University of Pittsburgh will conduct *Medical Genetics: 1993*, intended as a review for the examination of the American Board of Medical Genetics a month later. Local and visiting faculty will use lectures, workshops, and practice questions to review dysmorphology, quantitative genetics, and cytogenetic, biochemical, and molecular diagnostics, as applied to clinical and counselling cases. For information, contact Department of Conference Management, University of Pittsburgh Medical Center, Nese-Barkan Building, Fifth Floor, 3811 O'Hara Street, Pittsburgh, PA 15213-2593, USA. Tel: (412) 647–8232. Fax: (412) 647-8222.

NOTICE

European School of Medical Genetics

European School of Medical Genetics, 6th Course, Sestri Levante (Genoa), 28 March to 3 April 1993.
Directors Professor V A McKusick (Baltimore), Professor G Romeo (Genoa).
Faculty S Antonorakis, S Aymé, A Ballabio, R Balling, G Barbujani, C Bordignon, J Burn, J-J Cassiman, A Chakravarti, P Demant, M Devoto, M Goossens, L Heredero, W Lo, A Monaco, J Ott, M Pembrey, L Sandkuijl, G J van Ommen, J Weissenbach, P Scambler, A Schinzel, J Zonana.
Topics Introduction to Medical and Molecular Genetics, Mendelian and non-Mendelian Genetics, Human and Clinical Cytogenetics, Radiation Hybrids, FISH, Population Genetics, Molecular Genetics, Counselling, Screenings, Prenatal Diagnosis, Congenital Malformations, Model Systems.
Applications Send a CV, a brief description of your research interests, a letter of presentation, and a certificate of your knowledge of English to M Caterina Cogorno, Laboratory of Molecular Genetics, Istituto G Gaslini, 16148 Genova, Italy. Tel: +39/10/5636370-400, Fax +39/10/391254, E-mail: GENETICA@IGECUNIV.

Correction

In the article 'Attitudes towards prenatal diagnosis and carrier screening for cystic fibrosis among the parents of patients in a paediatric cystic fibrosis clinic' by Watson *et al* (*J Med Genet* 1992; **29**: 490–1), one of the questions was accidentally omitted from the questionnaire as published in the Appendix. The missing question (9) is "Do you think that introduction of screening to detect carriers in the population would be a good thing?", to which 92% answered yes.

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. *Short reports* or *Short communications* should in general not exceed 500 words, with one or two illustrations. Contributions may also be submitted as *Hypotheses* or *Technical notes*. 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.