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BOOK REVIEWS

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PCR Protocols—Current Methods and Applications. Methods in Molecular Biology vol 15. Ed Bruce A White. (Pp 392.) Totowa, NJ: Humana Press. 1993. ISBN 0-89603-244-2.

This book is the latest in the Methods in Molecular Biology series and it is aimed at anyone using the PCR technique in any applicable field of research. With less emphasis on specific examples and hints for modification of particular protocols, the book aims to provide easily adaptable frameworks to suit most PCR based experimental strategies. Specific examples are provided throughout the book but, unlike many texts, suggestions for troubleshooting and modification of techniques are plentiful as well. Subjects range from a basic introduction to PCR and primer design to the complexities of PCR mutagenesis, genomic footprinting, and identification of new members of gene families.

For the researcher with PCR experience, this book offers practical help with a large range of techniques and includes many useful hints for optimisation of the described protocols. However, for the novice, I would not recommend this book for a number of reasons. There has been no attempt made to group techniques by subject, so once past the introductory chapters there is little guidance as to the suitability of particular protocols for particular projects. This omission is compounded by the listing of key topics and techniques on the back cover without any references to the main text. The space taken up by the unreadable listing of all the chapter titles on the back cover, which is in small type with no gaps between titles, could have been better used with a brief index. The organisation of each chapter is also unfriendly because the materials, methods, and notes sections are separated. This means the reader frequently needs to flip between three pages to obtain the full details of a protocol. It is ironic that the book is comb bound to allow it to lie flat on the bench because the layout means that the book will spend little time in this position as the reader tries to piece together the whole technique.

This book is not an essential purchase for the PCR worker but it is a valuable addition to the field, with the caveat that the useful details are often less than easy to find, which reduces its utility. For the beginner, I would be happier to recommend other works on PCR which have a clearer, more concise layout for both the theory and practice of most of the commonly used PCR protocols.

ANDREW J WALLEY

The Molecular and Genetic Basis of Neurological Disease. Ed R N Rosenberg, S B Prusiner, S DiMauro, R L Barchi, L M Kunkel. (Pp 1023; £175.00.) London: Butterworth-Heinemann. 1993.

The editors describe the first edition of this book as being "at the cutting edge of the interface between clinical and basic neuroscience". This is reflected in their choices of contributors, all authorities in their fields and with high profiles in the author lists in current neurology/genetics journals. The chapters are, in general, comprehensive, well written, and more than adequately referenced. The book is divided into sections, each containing between one and five chapters.

Part I, chapter 1, begins gently with descriptions of nucleic acids and DNA structure and translation, and progresses rapidly (in 17 pages) through DNA markers, restriction enzymes, and polymorphisms to linkage, cloning, and gene therapy. One small criticism is the use of the term 'reverse genetics' where most would now use 'positional cloning'. In addition the section devoted to Lesch-Nyhan disease would seem superfluous in view of the full chapter coverage later.

Chapter 2 is a list of the chromosomal locations for genetic neurological diseases and genes of relevance to the nervous system. There are some minor omissions: tuberous sclerosis has entries at 9q34, 11q14, 12q22 (the last two are rather dubious localisations) but not chromosome 16 where there is a major locus. This is, however, mentioned in the chapter on TS. Similarly, there are no listings for myotonia congenita and Becker generalised myotonia which are associated with the skeletal muscle chloride channel at 7q32-qter. Some entries list the disorder alongside the gene product but this is not universal; dystrophin does not appear with Duchenne and Becker muscular dystrophies. Chapter 3 gives a small (three pages) taste of linkage analysis, computer programs for such, and the problem of genetic heterogeneity. It is perhaps a little too short for a reader with no previous knowledge of this field.

The disease specific chapters which follow contain many highlights and very few disappointments. The chapters on metabolic disorders admirably cover the clinical, biochemical, and molecular features of the appropriate diseases. In addition they offer therapeutic protocols where available, or discuss potentials for therapy. Part XII contains a single chapter; a scholarly multiauthor review of the prion diseases of humans and animals. Prusiner and colleagues also provide ample scope for further reading on this fascinating topic with 311 references.

Part XIII contains reviews of the muscle dystrophies and, with the exception of the chapter on FSH/limb-girdle dystrophies which contains less than two pages of text, this section amply summarises the salient clinical and molecular features of these disorders. Roses (chapter 41) has been able to incorporate some of the rapidly appearing new data on the unstable trinucleotide repeat sequence associated with myotonic dystrophy and this chapter is well referenced up to 1992. At this time it was known that expansion of another trinucleotide repeat sequence, this time in the androgen receptor gene, was the causal mechanism of bulbospinal neuronopathy (Kennedy's syndrome). There is no mention of this, either in this chapter or (that this reviewer could find) elsewhere in the book.

Part XIV contains, again, a single contribution. DiMauro's review of mitochondrial disorders is comprehensive, clearly written, well supplemented with explanatory diagrams, and excellently referenced. Part XV deals with 'degenerative disorders' and begins with a chapter on hereditary ataxias. Unfortunately this was a little disappointing as over half of this chapter reviews disorders given more comprehensive coverage elsewhere in the text. Part XIX (neuronopathies and neuropathies) contains limited clinical information, the discussions focusing on gene mapping (for spinal muscular atrophy), mutations (transthyretin and amyloid neuropathies), and peripheral nerve physiology (axonal neuropathies). The review of the genetic epilepsies (part XX/chapter 63) is particularly thorough, including a description of the epileptic Mongolian gerbil! It also includes useful guidelines on risk estimates and prognosis for use in counselling relatives of epileptic persons. The final chapters (gene therapy and consequences of gene mapping) conclude this volume with general discussions on likely future developments in the field of neurogenetic diseases, although specific examples are given for replacement therapy in animal models. The reviewer finds it surprising that there is no mention of the possible societal and political consequences of the Human Genome Project.

Is this the neurology equivalent of *The Metabolic Basis of Inherited Disease*? The answer must be 'yes'. The criticisms above are minor compared with the wealth of information provided overall and this volume will certainly fulfil its goal in being of use to clinicians caring for patients and for scientific investigators in this field. Every genetic/neurology unit should have access to this volume but most people will want their own copy and anyone with a colleague visiting North America would do well to persuade him/her to add the extra weight to their return luggage as the US price equates to £150.

JOHN C MACMILLAN

NOTICE

European School of Medical Genetics: 7th Course

This course will be held in Sestri Levante, Genoa, 20 to 26 March, 1994. Directors: Professor V A McKusick (Baltimore), Professor G Romeo (Genoa). Faculty: S Aymé, A Ballabio, M Baraitser, G Barbujani, B Brambati, J-J Cassiman, A Chakravarti, M Devoto, M Gershon, P Goodfellow, D Housman, G Jackson, R Kapur, A Monaco, B Müller, V Pachnis, M Pembrey, B Ponder, G J van Ommen, A Schinzel, G Simoni, S M Tilgham. Topics: Introduction to human molecular genetics, linkage analysis, mouse genetics, population genetics, cancer genetics, physical mapping, positional cloning, candidate genes, clinical dysmorphology, genetic counselling, community genetic services. Registration fee: 350 000 Italian lire. Applications: send your CV, a brief description of your research interests, a letter of presentation (if you wish to present a clinical case during an evening session and to apply for a travel fellowship in case there are some available, please state it clearly in your cover letter), 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.

Correction

In the paper 'A syndrome of insulin resistance resembling leprechaunism in five sibs of consanguineous parents' (*J Med Genet* 1993;**30**:470–5), fig 2B is of IV.1 and fig 3B of IV.5.

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. If requested, authors shall produce the data upon which the manuscript is based for examination by the Editor. Guidance on length can be obtained from studying the Journal. Case and family reports may be submitted as *Brief papers*. *Short reports* should in general not exceed 500 words, with one or two illustrations, and the text should be continuous with no headings. An abstract should be provided for all papers. 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.

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.

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.

GUIDELINES FOR SUBMISSION OF REVISED PAPERS

A revised manuscript should be returned within two months. Manuscripts returned after two months will be treated as new papers. When submitting a revised manuscript please ensure you enclose three copies of this and one copy of the original manuscript.