
Journal of

MEDICAL GENETICS

Editor: Peter S Harper

North American Editor: P Michael Conneally (Indianapolis)

Reviews Editor: Rodney Harris (Manchester)

Cytogenetics Editor: Patricia A Jacobs (Salisbury)

Molecular Genetics Editor: Ann Harris (Oxford)

Technical Editor: Clare Henderson

EDITORIAL COMMITTEE

V Baranov (St Petersburg)

D Timothy Bishop (Leeds)

A Cao (Cagliari)

Ann C Chandley (Edinburgh)

David R Cox (San Francisco)

A E Czeizel (Hungary)

Dian Donnai (Manchester)

J P Fryns (Leuven)

T Gedde-Dahl Jr (Tromsø)

P N Goodfellow (London)

Karl-Heinz Grzeschik (Marburg)

Judith G Hall (Vancouver)

A E Harding (London)

M R Hayden (Vancouver)

Thaddeus E Kelly (Charlottesville)

P McGuffin (Cardiff)

Victor A McKusick (Baltimore)

Jean-Louis Mandel (Strasbourg)

T Mazurczak (Warsaw)

Margareta Mikkelsen (Copenhagen)

Andrew P Read (Manchester)

A Schinzel (Zurich)

Grant R Sutherland (Adelaide)

G J B van Ommen (Leiden)

Tessa Webb (Birmingham)

Robin M Winter (London)

I D Young (Nottingham)

Y T Zeng (Shanghai)

Editor,

British Medical Journal

NOTICE TO ADVERTISERS

Applications for advertising space and rates should be made to the Advertisement Manager, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR.

NOTICE TO SUBSCRIBERS

Journal of Medical Genetics is published monthly. The annual subscription rates are £163.00 (US \$277.00). Orders should be sent to The Subscription Manager, *Journal of Medical Genetics*, BMA House, Tavistock Square, London WC1H 9JR. Orders can also be placed with any leading subscription agent or bookseller. (For the convenience of readers in the USA subscription orders with or without payment may be sent to British Medical Journal, Box 560B, Kennebunkport, Maine 04046, USA. All enquiries, however, must be addressed to the Publisher in London.) Subscriptions may be paid by Access, Visa, or

American Express by quoting on the order the credit or charge card preferred together with the appropriate personal account number and the expiry date of the card. All enquiries regarding air mail rates and single copies already published should be addressed to the Publisher in London. Second class postage paid, Rahway N J Postmaster: send address changes to *Journal of Medical Genetics*, c/o Mercury Airfreight International Ltd Inc, 2323 Randolph Avenue, Avenel, NJ 07001, USA.

COPYRIGHT © 1993 by *Journal of Medical Genetics*. All Rights Reserved. No part of their publication may be reproduced, stored in a retrieval system, or transmitted in any form or by any means, electronic, mechanical, photocopying, recording or otherwise, without the prior permission of *Journal of Medical Genetics*.

ISSN 0022-2593

Published by the BMJ Publishing Group, BMA House, Tavistock Square, London WC1H 9JR, and printed in England by Latimer Trend & Company Ltd, Plymouth.

BOOK REVIEWS

If you wish to order or require further information regarding the titles reviewed here, please write to or telephone the BMJ Bookshop, PO Box 295, London WC1H 9JR. Tel 071 383 6244. Fax 071 383 6662. Books are supplied post free in the UK and for BFPO addresses. Overseas customers should add 15% 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. (The price and availability are occasionally subject to revision by the Publishers.)

Our Genetic Future: The Science and Ethics of Genetic Technology. (£7.99.) London: British Medical Association. 1992.

I found it difficult to review this book, perhaps reflecting the difficulty that must have been experienced by the committee that wrote it. This volume is the report of a BMA Working Party charged to examine the implications of genetic engineering, and consists of 236 pages of text with a few black and white diagrams, plus references and a glossary. In the first eight chapters and 135 pages it sets out to inform the lay reader about mendelian genetics, molecular genetics, and the application of genetic engineering to plant and animal breeding, to the production of pharmaceutical compounds and vaccines, and to prenatal and carrier genetic screening and diagnosis. On the one hand, there is too much information for the layman, and the illustrations are not of sufficient number or quality to compensate; there are much better and more attractive primers available. On the other hand, this material is not really very helpful to those who are already familiar with the field.

The remaining six chapters and 101 pages are most useful. Chapter 9 reviews the current laws and guidelines concerning gene manipulation in Britain, Europe, and the USA. Chapter 11 discusses the possible benefits and problems associated with genetic modification of plants, animals, and micro-organisms; it contains one of the (too) few critical passages in the book, when explaining how the Green Revolution of super yielding cereal varieties resulted in disaster for some Third World agricultural communities. It follows this by pointing out that commercial interests may conflict with interests of mankind in such areas as the maintenance of genetic diversity, but its recommendations are vague. Chapter 11 presents some of the arguments surrounding the patenting of genetically modified organisms, and presents the contrasting situations in Europe and the USA. Chapter 12 briefly discusses the application of gene manipulation to pharmaceuticals, and then presents the welcome, and surprisingly radical, case for demanding that a fourth criterion (or hurdle) be met before a new drug is introduced. In addition to the criteria of safety, efficacy, and quality, there should be a social criterion: the product should be 'needed', and its likely social impact should be beneficial. If this criterion were applied in a wider context, then genetics and the rest of medicine, and indeed our whole society, would be transformed, but this avenue was not pursued.

The next two chapters cover in 45 pages the implications of the application of genetic

modification to people and of the human genome project. This material should have been the core of the book, instead of a mere tail. Chapter 13 examines gene therapy, genetic screening, confidentiality (on which a rather weak stance is adopted), occupational genetic screening, insurance, and DNA fingerprinting. Chapter 14 examines the patenting or copyrighting of human DNA sequences, and explores the potential conflict between commercial interests and medical need for the public health. Finally, there is a summary and a generally sensible and worthy set of recommendations.

I was disappointed at the tenor of the volume, which is for the most part bland and uncritical; while the possible technical and administrative problems arising from genetic manipulation are discussed, the broader issues receive inadequate attention. Thus, some of the psychosocial issues arising from carrier screening for recessive disease are outlined, but far too little attention is paid to this area if it is considered that the report comes from a medical body. The problems that may arise if the attention and resources of health services, and of society at large, concentrate on genetic explanations of ill health (geneticisation) are not addressed. The final few chapters of this report do provide an interesting review of selected topics, and I would like to see the summary and recommendations being widely discussed; however, I cannot recommend the volume as a whole to any particular readership.

ANGUS CLARKE

Practical Molecular Virology. Methods in Molecular Biology volume 8. Ed Mary K L Collins. (Pp 330.) London: Humana Press. 1991.

My ideal of a laboratory methods book is one that gives a succinct, step by step summary of the practical techniques, with a short section on the applicability and the limitations of each technique, and a brief troubleshooting guide. I also like to see remarks on the relative importance of the steps, so that I can judge which are most likely to cause difficulties, and which can be left out. It should be written by a single author, or tightly edited so as to give a uniform style. The famous 'Maniatis' (now Sambrook *et al*) inevitably springs to mind as a first rate example in all of these aspects. This book largely meets these criteria, but the coverage of some of the topics is uneven, and I suggest that you look closely at the relevant chapters to see how well the technique you need is covered.

The subtitle of the book is 'Viral Vectors and Gene Expression', and this clearly shows that the scope of the book is not as wide as the main title suggests. There are 28 chapters, primarily on the use of viruses as vectors for foreign gene expression, interspersed with more general chapters on techniques such as PCR and syncytial assays. There is a heavy emphasis on retroviral vectors (eight chapters), which is unsurprising in the view of the authors and the editor: there are also three chapters on baculoviruses, two each on Herpes simplex, Epstein-Barr virus, vaccinia virus, poliovirus, and virus-like particles in yeast, and one each on adenoviruses and papillomaviruses. The treatment of retroviral vectors includes their use to mark cell lineages and as insertional mutagens.

Background information to the methods is usually well treated, particularly in the case

of the excellent introductory chapters on retroviruses, and the authors have taken trouble to indicate whether a given vector is likely to be suitable for your purposes. Almost inevitably in a multi-author volume, however, the level of detail is highly variable in both the background information and the actual methods. This is reflected in the number of references at the end of the chapters, which varies from five to over 100. A small number of the chapters are, as a result, so general as to be almost useless as a practical guide.

On the back cover of the book, the publisher writes "Humana's trend-setting series... 'Hands-on' techniques that everyone can use!" This surely applies to most of the techniques described, but in the case of certain notoriously exacting and fickle techniques, such as the production of vesicular stomatitis virus pseudotypes, you would be well advised to consult someone, preferably the author of these chapters, before embarking on them.

To summarise, the book is very likely to be useful if you intend to use the viruses covered as vectors, especially if you want to use retroviruses, but don't expect a comprehensive coverage of molecular techniques, and consider carefully before planning to use VSV pseudotypes!

CHARLES R M BANGHAM

NOTICES

The New York Academy of Sciences

The New York Academy of Sciences is sponsoring the following symposia and conferences. 'Microbial Pathogenesis and Immune Response' on 8 to 11 September 1993 at Grosvenor Resort, Walt Disney World Village, Orlando, Florida, USA.

'Fetal Cells in Maternal Blood: Prospects for Noninvasive Prenatal Diagnosis' on 27 to 29 September 1993 at Crystal Gateway Marriott, Arlington, Virginia, USA.

'DNA Damage: Effects on DNA Structure and Protein Recognition' on 31 July to 4 August 1993 at Radisson Hotel, Burlington, Vermont, USA.

'DNA: The Double Helix. Forty Years: Perspective and Prospective' on 13 to 16 October 1993 at Chicago Marriott, Downtown Chicago, Illinois, USA. (Cosponsors The University of Illinois at Chicago and Green College, Oxford University.)

For further information contact: Conference Department, New York Academy of Sciences, 2 East 63rd Street, New York, NY 10021, USA. Tel (212) 838-0230, fax (212) 838-5640.

International Symposium on Neurofibromatosis

In collaboration with Professor Jean-Pierre Fryns, University of Leuven and Professor Claude Stoll, University of Strasbourg, the National Neurofibromatosis Foundation and the International Neurofibromatosis Association are organising an International Symposium on Neurofibromatosis to be held on 11 and 12 September 1993 in Strasbourg, France, as an adjunct meeting of the IV European Dysmorphology Meeting. For further information contact Sherland Peterson-Ortiz, National Neurofibromatosis Foundation, 141 Fifth Avenue, Suite 7-S, New York, NY 10010-7105, USA. Fax: (212) 529-6094, tel: (212) 460-8980.

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*. 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.