
Journal of

MEDICAL GENETICS

Editor: Peter S Harper
North American Editor: P Michael Conneally
Cytogenetics Editor: Patricia A Jacobs
Molecular Genetics Editor: Kay E Davies
Reviews Editor: Rodney Harris
Technical Editor: Clare Henderson

EDITORIAL COMMITTEE

V Baranov (Petrograd)
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 (Tromsö)
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 £148.00 (US \$250.00). 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 © 1992 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

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.

Genetic Data Analysis. Methods for Discrete Population Genetic Data. Bruce S Weir. (Pp 377.) Sunderland, Mass: Sinauer Associates. 1991.

The subject of this book is further specified in the preface as "methods for traditional analyses of Hardy-Weinberg and linkage disequilibrium and methods for characterizing population structure and estimating genetic distance. It also introduces the reader to some of the many methods of constructing evolutionary trees and to the difficult problems of aligning and comparing sequences". This is a warning not to expect segregation, linkage, association, mutation, or gene mapping, but does not prepare the reader for several other limitations. First, there has been no attempt to define basic concepts. For example, the first paragraph states that mendelising units would at one time have been called genes. The sophisticated reader knows that the opposite is true, while the naive but intelligent student will suppose that molecular biologists talk to each other about 'mendelising units' and will wonder what gene mappers map. Secondly, the object of many analyses is obscure. What biological principles have been clarified by variance analyses of codominant phenotype systems in arbitrarily stratified populations, what court would accept a paternity index based on 'prior belief', and have estimates of viability selection that neglect linkage disequilibrium, reproductive performance, and generation time seen any use in the last 20 years? Thirdly, few mathematical results are derived and the reader must take a great deal on faith. Finally, within its limited scope this book has remarkable omissions. All phenotypes are restricted to fully penetrant genes, usually codominant, and gene interactions are not considered. Population structure is discussed without reference to inbreeding or outcrossing effects, isonymy, kinship, genealogy, migration, isolation by distance, or observations in any population. Many papers of the North Carolina group are cited, but none of Cotterman, Malecot, Crow, or Jacquard.

Despite these limitations this book has several advantages. It contains a number of useful references from the last few years and provides a good introduction to the work of Cockerham and Weir and to sequence analysis. The variety of topics covered is helpful for critical readers already familiar with other aspects of genetic analysis, which is evolving so rapidly that a new book is always welcome.

NEWTON E MORTON

Genetic Disorders of the Skin. Ed Joseph C Alper. (Pp 378; £106.00.) St Louis: Mosby Year Book, Inc. 1991.

The 20 chapters in this book, which are written entirely by North American contributors, most of whom are dermatologists or paediatricians, effectively cover the increasingly complex field of heritable skin diseases or 'genodermatoses'. The organisation of the chapters seems partly logical and partly arbitrary. The first chapter on principles of genetics is a brave attempt to cram a large amount of information, possibly too much, into 29 pages. As a result, the diagram showing the morbid anatomy of the human genome on two pages (25 × 17 cm) is almost indecipherable. There are chapters on development of the skin, genetic counselling, chromosome disorders, and biochemical disorders. Other chapters focus on more specific subjects, such as hereditary bullous disorders, keratinising disorders, and connective tissue disorders. These chapters include sections on clinical features, laboratory investigations, diagnosis, prenatal diagnosis, and treatment. Certain chapters, such as the one on diseases associated with DNA and chromosomal instability, are particularly good and comprehensive for a book of this size, whereas the chapter on neurofibromatosis and Albright's syndrome is disappointingly brief. The last chapter on legal aspects of hereditary cutaneous diseases is written by two lawyers and seems an odd inclusion for a book of this sort. The ethical aspects of genetic diagnosis and screening are hardly addressed: this contribution is much more concerned with the problems of malpractice and litigation.

The strength of the book is that it is currently the most comprehensive text on the subject of genetic skin disorders. Its weakness is the lack of uniformity between the organisation of chapters, even allowing for the varied writing styles of the different authors. The overlap between chapters also leads to difficulty in tracking down essential information. For example, the photograph of nail changes in dyskeratosis congenita is included in the section on genodermatoses with malignant potential instead of that on genetic nail disorders. A large number of black and white clinical photographs are of poor general quality and even out of focus.

This book will provide a useful reference source for dermatologists, paediatricians, and clinical geneticists interested in the management of genetic disorders of the skin, but is likely to be too expensive for most personal bookshelves.

R A J EADY

Cystic Fibrosis. Ed Richard D Moss. (Pp 251; £75.60.) Bristol: Humana Press. 1991.

This book consists of 12 reviews of aspects of cystic fibrosis. The authors are chiefly from the United States. The reviews range from details of the CF gene and the cystic fibrosis transmembrane conductance regulator protein to clinically pertinent problems for physicians currently managing cystic fibrosis lung disease. The focus of this book is on the pathophysiology of cystic fibrosis lung disease with contributions on the relationship between atopy and cystic fibrosis, infection and immunity to *Pseudomonas* and other bacteria, viral infection, and heart-lung transplantation for end stage lung disease. Unlike

other works there are reviews of the role of virus infection, fungal infection, particularly allergic bronchopulmonary aspergillosis, and of infection of the sinuses and upper airways.

The review on infection with *Pseudomonas* species pays surprisingly little attention to infection with *P cepacia*, which is disappointing because this difficult infection in adult patients has been widely discussed in North American respiratory publications. Other reviews, such as the role of viruses in the exacerbation of respiratory symptoms, highlights the current lack of knowledge and the inadequacy of previous studies in this area. Overall these reviews lead one to the conclusion that much of the work carried out in the area of lung infection in cystic fibrosis has been in small numbers of patients in single centres and cannot hope to answer the questions being posed adequately. It argues very much for what is now occurring in the United States; multicentre studies in statistically relevant population sizes where allowance for the wide range of severity of disease in such patients can be made.

This book has many weaknesses and some strengths. It is not a book for the general reader or the clinician purely concerned with the management of respiratory problems in cystic fibrosis. It is possibly a book for the enthusiast and researcher working in this area and provides some useful overviews of a range of problems associated with pulmonary disease in cystic fibrosis.

DENNIS J SHALE

NOTICES

International Conference on Molecular Biology of Genetic Diseases

A conference on modern aspects of the diagnosis, pathophysiology, and treatment of genetic disease will be held on 4 to 7 October 1992 at the Jin Jiang Hotel in Shanghai, China under the auspices of the Shanghai Children's Hospital. Sessions will include gene transfer and transgenic animals, genomics and reverse genetics, globin genes and haemoglobinopathies, inborn errors of metabolism, gene therapy, and the application of new procedures in clinical genetics. Presentations will be by lecture and poster. For more information about the conference, please contact Conference Secretariat, c/o Dr Edward R B McCabe, Institute for Molecular Genetics, Baylor College of Medicine, One Baylor Plaza, Houston, Texas 77030, USA. Tel: (713) 798-5820, Fax: (713) 798-5881.

International Congress on Improving Birth Quality and Child Upbringing

This Congress, jointly sponsored by the Ministry of Public Health, the Chinese Association for Improving Birth Quality and Child Upbringing, and the China International Conference Center for Science and Technology, will be held in Beijing from 24 to 27 May 1992. For further information contact Ms Jin Fang, ICIBQCU, Congress Secretariat, PO Box 300, CICCST, Beijing 100086, China. Tel: 011-86-1 8313335, Fax: 011-86-1 8316091.