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BOOK REVIEWS

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Muscular Dystrophy—The Facts. A E H Emery. (Pp 135; £7.99.) Oxford: Oxford University Press. 1994. ISBN 0-19-262449-0.

This small paperback book is an excellent addition to an excellent series of some two dozen books published by Oxford University Press, aimed at patients and their relatives. Other neurological titles in "The Facts" series include Parkinson's disease and multiple sclerosis, and for some years I have recommended them to most of my patients with these disorders and received appreciative comments. Titles of interest to geneticists include cystic fibrosis and Down syndrome.

Alan Emery needs no introduction to the readers of this journal and it is difficult to think of a more appropriate author for such a text. The style which makes him such a popular lecturer and companion shines through. For example, I had always believed that the term lordosis was derived from the Greek word lordos, meaning bent, but now know that the term arose because of the resemblance of the posture to the gait of a peer of the realm! The book is aimed at a lay audience and with the help of a useful glossary it is certainly pitched at the right level. As well as its intended audience I wholeheartedly commend it to all medical and allied staff involved in the management of patients with muscular dystrophy.

The main emphasis of the book is medical problems and their management. The meaning of the term muscular dystrophy is discussed, followed by chapters on confirmation of the diagnosis and on outlining the main clinical features of the different types of muscular dystrophy. Not surprisingly, the chapter on inheritance and genetic counselling is particularly good. Although stated to be a lesser aim of the book, there are useful chapters on practical issues including living with muscular dystrophy, education, and employment, and the range of professional and voluntary support services available, with extensive recommendations for further reading and a list of addresses of muscular dystrophy associations throughout the world.

What criticisms I have are few and trivial. When discussing muscle biopsy it is stated that general anaesthesia is preferred to local. While perhaps true in early childhood this is obviously not the case in adults. It may seem cheeky to criticise the section on Emery–Dreifuss muscular dystrophy, but as Emery has himself previously discussed it is probably more helpful to think in terms of an Emery–Dreifuss syndrome. Thus, a similar phenotype but with autosomal dominant inheritance is also recognised. This section will in any case need revision before the next edition, following the recent identification of the gene involved in the X linked form, and its protein product emerin. A final section of the book, Living with Muscular Dystrophy, contains just photographs and captions. I didn't feel that this was useful and some of the captions verged on the condescending. I must emphasise that my criticisms are minor. There are no factual errors and I have no doubt that this book will be greatly appreciated not only by its intended audience of patients and their families, but also by their carers.

DAVID HILTON-JONES

Current Protocols in Human Genetics. Editors N C Dracopoli *et al.* Series Editor A L Boyle. New York: Green Publishing Associates and Wiley. 1994.

This is another excellent addition to the Current Protocols series using the now familiar format that has made the "Red Book" such a valuable reference work.

Chapters are generally well written and give excellent and detailed insights into the protocols they describe. The common problems and traps are highlighted in all protocols. Valuable safety data are also given and stressed. One potential criticism is that background information about the protocols should come before the detailed techniques. This gives you a better insight into the procedure when reading through the manual in a sequential manner.

Obviously people differ in their choice of particular techniques. For example, in unit 2.5.1, Methods in Genotyping, a lot of stress is placed on labelling SSLPs directly. This is expensive in terms of radioactivity used, around 10 μCi per 96 well plate and probably not so good for the person doing the work. Another method of visualising SSLP reaction products is to do the PCR without any radioactivity then blot the acrylamide gel on to Hybond N+. This is then probed with a radioactive oligonucleotide against the repeat used. Under these conditions a total of 10^5–10^6 counts is used per hybridisation and the hybridisation solution with the radioactive probe can be reused up to five times. Hence, excessive quantities of P^{32} are avoided and the radioactivity is contained.

There is an acknowledged need to demystify much of cytogenetic methodology. The cytogenetics chapters go some way towards fulfilling this need, by providing clear and concise methods for a range of cytogenetic techniques together with their scientific basis.

Chapter 4 includes methods for the preparation of metaphase cells from peripheral blood lymphocytes as well as an exhaustive section on banding techniques and their uses. In addition to these conventional cytogenetic approaches, new molecular cytogenetic techniques are presented including in situ hybridisation with fluorescent, enzymatic, and radioisotopic detection strategies. The section on microscopy provides a more than adequate overview of bright field and fluorescent microscopy as well as an introduction to image analysis for karyotyping and FISH.

In 4.1, Peripheral Blood Culture, a method for standard culture and harvest as well as two methods for high resolution chromosomes are given. The methotrexate synchronisation method is an unusual choice as it results in a high percentage of cells with chromosome breakage. Several other methods for obtaining high resolution chromosomes are more widely used (at least in Europe), including synchronisation with excess thymidine and BUdR incorporation.

A full two pages are devoted to a very detailed description of one method for slide making. Although they admit that there are many different methods for slide making, the method given is quite a difficult one. More useful is a discussion of the appearance of well spread metaphases under phase contrast and how to alter slide making procedures according to the changing environment in the laboratory.

No cytogenetic methods are given for fragile X detection: with the discovery of more fragile sites at Xq28 (FRAXE, FRAXF) which are not detectable by molecular analysis for FRAXA, it may be premature to discard cytogenetic analysis for fragile sites.

Many of the methods presented are more widely used in the USA than in Europe; the American Cytogenetic Technologist's Laboratory Manual is extensively quoted. Similarly, the quality control/assurance regulations quoted are specifically applicable to US laboratories. However, there is a very useful section on the principles which should be adhered to in running a diagnostic cytogenetics service and includes tips which apply equally well to research settings. Similarly, the recommendations for the number of cells to analyse and karyotype are very helpful as guidelines where no local rules exist.

In keeping with the Current Protocols series, *Current Protocols in Genetics* provides an informative guide to cytogenetic methods which can be easily updated by the addition of supplementary sections. However, the inclusion of fig 4.5.2 is a little confusing as the section on extended DNA preparations (unit 4.5) is not included.

In chapter 5, Large-Insert Cloning and Analysis, the authors do not seem to stress the idea of producing a cosmid or λ phage library from the total yeast DNA and screening with left and right arm probes to obtain the ends of the YAC followed by screening with total human DNA to obtain sections of the YAC insert. The production of total yeast libraries and screening for YAC ends is effective and rapid. The probes that are derived from λ phage or cosmid library are always big enough to map the ends of the YAC very efficiently, either on hybrid panels or by FISH.

The chapters on defining genes involved in disease and then defining mutations within them are comprehensive, well laid out, and the background information is informative without being boring. The protocols at the heart of these chapters are relatively easy to follow, though continual references to methods located elsewhere in the book can be tedious, but this is an understandable concession to keeping this volume's size under control.

It should also be noted that the protocol describing exon trapping in chapter 6 only recommends a Gibco/BRL vector. USB also make a suitable vector for such studies and recommendations from authors who work for Life Technologies, of which Gibco is part, give an unwanted bias to this section which is in marked contrast to the rest of this excellent manual.

Chapter 8 contains sections on the culture and preparation of chromosomes from chorionic villus samples (CVS), amniotic fluid,

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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.

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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.

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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.

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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.

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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 the 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.

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