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 μ Ci per 96 well plate and probably not so good for the person doing the work. Another method of visualising SSCP 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,

and fetal tissue. The overview of clinical cytogenetics provides a very good summary of the indications for pre- and postnatal cytogenetic studies and the means by which these can be achieved in a clinical setting. In addition, a brief guide to the major chromosome abnormalities is given. The background information on CVS and the relative advantages and pitfalls of direct and culture methods is well presented. The chapters on amniotic fluid and fetal tissue analysis provide an excellent and informative review of these subjects, with a wide range of techniques that should suit all tastes. Choice of in situ or flask harvesting methods is given and the different requirements for analysis and the detection of mosaicism from these two different types of harvest is discussed in detail.

In all cases the sections devoted to background information, troubleshooting, and time consideration provide a helpful adjunct to the methods.

Current Protocols in Human Genetics can be wholeheartedly recommended to all workers in the human genetics field with a basic grounding in molecular biology, but you will have to buy *Current Protocols in Molecular Biology* if you are not to be left searching for basic protocols essential for your research.

L KEARNEY, A WALLEY,
C WARD, A HARRIS

Anthropometry: the Individual and the Population. Cambridge Studies in Biological Anthropology. Editors S J Ulijaszek, C G N Mascie-Taylor. (Pp xiii + 213). Cambridge: Cambridge University Press. 1994.

This is the fourteenth book in the *Cambridge Series in Biological Anthropology*, a series which has distinguished itself by providing an excellent number of publications both in biological anthropology per se and in closely related fields of study.

For a considerable period, anthropometry constituted the most important of the investigatory techniques used by physical anthropologists, both to examine the morphological relationship of extinct and extant populations, and also to examine so called racial variability. With the development of human population genetic studies in the 20th century it was increasingly assumed that, in many contexts, anthropometry was rather old fashioned, and it became somewhat discredited.

However, this volume abundantly demonstrates that anthropometric techniques, procedures, and results all have value in a number of somewhat distinctive contexts. Many of these are dealt with in the chapters of the present volume. There is a succinct and valuable introduction by Lasker, in which he outlines the changes in research paradigms over the years. The 11 remaining chapters in turn are devoted to the following topics: asymmetry and growth, intra- and inter-observer error in anthropometric measurement, statistical issues in anthropometry, other aspects of human growth, variability in adult body size, anthropometry and body composition, physical performance and motor fitness, and, lastly, anthropometry in the US armed forces. It is evident that a wide range of diverse topics is covered in this volume, and it is, in my view, appropriate that the multi-faceted nature and uses of anthropometry should thus be elucidated.

Those with an interest in any one of the

components listed above will find useful, up to date, state of the art material in this book. The chapters are, on the whole, well and fully referenced, and these can serve as starting points for more detailed, particular studies. The diversity of the approaches is such that the book may readily be commended to many specialists. It is a distinguished addition to the volumes already produced in this particular series.

ERIC SUNDERLAND

Chromosome Techniques—A Manual. A K Sharma, A Sharma. (Pp 368; £55.00) Chur, Switzerland: Harwood, 1994.

The authors' intention with this book is to provide a comprehensive set of protocols enabling the researcher or student to obtain and examine chromosome preparations from just about any organism from a dinoflagellate to a mammal. It covers methods for fixation, staining, banding, plant and animal tissue culture, cell fusion, autoradiography, monitoring (sic) for environmental toxicants, in situ hybridisation, and the underlying DNA techniques required for such methods, immunofluorescence and antibody labelling. Approaches to chromosome examination by a range of microscopic techniques are discussed, including phase and interference, confocal, fluorescence, and electron microscopy. In addition, there is some discussion of the theoretical principles relating to the preparatory methods and microscopy. This book contains a wealth of information, and it is certainly enlightening to read of the myriad different ways in which chromosomes may be prepared, manipulated, and examined; despite this, to fulfil its intention in a mere 368 pages is a tall order.

Reviewing this all encompassing volume from the standpoint of a clinical cytogeneticist, my approach has been to determine whether those methods with which I myself am more familiar are dealt with adequately, and I have had to make some assumptions that other topics are covered to a similar standard.

I found the book to be an unusual blend of ancient and modern, with methods apparently often taken directly from laboratory notes and working protocols over many years. Just one example of this blend concerns a description of microphotography being undertaken on large format plate cameras, reference to which occurs but a few pages before an outline of computerised karyotyping and image analysis. It is a pity that, having discussed the production of hard copy images, a book dealing with an essentially visual science should contain not one single illustration.

The methods suggested for mammalian tissue culture mostly belong in the archives (along with that plate camera), certainly as far as the clinical cytogenetics laboratory is concerned. Separating leucocytes for short term culture was out of date 25 years ago, and the practice of embedding fibroblast explants in chick embryo extract and cockerel plasma is similarly antiquated. Inclusion of historically important methods would be fine as long as more applicable up to date information were provided also. However, no mention at all is made of the current, much more straightforward, approach to tissue culture made possible by modern equipment, synthetic growth factor supplemented media, and years of experience; nor of the production

of elongated chromosomes for high resolution analysis by synchronisation or intercalating agents.

To use this volume as a bench top recipe book would be difficult; the protocols are not easily followed, laid out as note form text rather than lists, not in any consistent format, and not always in the most logical order. To find one's way around a technical manual of this sort requires an adequate index. Unfortunately, the index is incomplete to the extent that cross reference between sections is impossible. For example, while Leishman stain is recommended in the method for trypsin G banding it appears neither in the index nor in the chapter on stains. Esoteric ingredients such as Abopon, Euparal, and Alcian blue appear seemingly at random without proper explanation of their sources or components. I encountered difficulties trying to track down protocols using the fluorescent dye acridine orange, where one important staining method, referred to in the text, is not indexed. Although references are appended at the end of the book they are incomplete; for example trypsin banding is described without acknowledgement of Seabright, counterstain enhanced fluorescence without mention of Schweizer. References are not systematically given in the text, so that referral back to the original source may not be possible. Readers not familiar with certain abbreviations will find the inconsistencies and lack of explanation of the abbreviations frustrating. Bromodeoxyuridine is given variously as BUdR, BrdU, and BrUrd. There is also disparity in the units used to describe concentrations: three consecutive methods give the concentration of the fluorescent dye Hoechst 33258 as a percentage, in moles, and in micrograms per millilitre respectively.

Some up to date methods are to be found in the latter part of the book, dealing with microdissection, in situ hybridisation, and special molecular techniques, although in this rapidly developing field the inclusion of some superseded methods is to be expected. Again, however, the incomplete indexing and cross referencing mean that only with difficulty could the protocols be attempted without extensive previous experience.

I have to admit that there are many topics included in this book on which I am not qualified to comment, but then few readers of *Journal of Medical Genetics* will want to culture pollen or make EM preparations from slime moulds. Basically, I believe that the authors have tried to cover too wide a range of methods and, while it may be of interest to the student as a resumé of what can be done with chromosomes and DNA, the book is not a practically useful bench manual. The scientist would be better advised purchasing two or three specialised texts, focusing in detail upon up to date information concerning specific topics. As far as medical geneticists are concerned, there is unfortunately little of practical value to recommend this volume.

R T HOWELL

Identification of Transcribed Sequences. Editors U Hochgeschwender, K Gardiner. (Pp 304; £89.00.) New York: Plenum Press, 1994. ISBN 0-306-44835-1.

One of the major challenges facing the Human Genome Mapping Project is how to