in families with IP may, however, be fruitful in the search for the gene.

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BOOK REVIEWS

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The A–Z Reference Book of Syndromes and Inherited Disorders. 2nd ed. Patricia Gilbert. (Pp 378; £15.99 pb.) London: Chapman & Hall. 1995. ISBN 0-412-64120-8.

The diagnosis of a rare condition in a child understandably raises many questions for that child's parents and relatives. Such queries are usually posed to the paediatrician or GP caring for the child. However, there are a wide range of health professionals involved in the care of the child who need information about a condition, both for themselves, and to answer the questions inevitably also posed to them. The problems of "how did it happen?", "can you treat it?", "will it happen again?", and many more are well addressed in this reference book.

Dr Gilbert has produced an expanded second edition of her book, designed to inform health professionals and families after a syndrome diagnosis has been made. It has been specifically written in non-technical language, aimed at a general readership, who may not have a broad medical knowledge. Twenty new syndromes have been included at the request of readers, giving a total of 90 conditions, the majority of which are genetic. This is assisted by a clear appendix containing a good review of basic genetics by Peter Farndon. A useful glossary is also included. There is an alphabetical listing of each syndrome, with 1000 words or more per entry describing its incidence, history, causation, characteristics, management, and future developments. The descriptions show that Dr Gilbert has had extensive first hand experience in the care and management of children with rare disorders. The inclusion of the addresses of support group associations, Contact-A-Family, and the UK clinical genetics centres also shows her understanding of parents' needs. The language used for the most part is clear and simple to understand.

No such book could aim to cover all the rare syndromes. The author states that she can only cover a small number of syndromes. Her book is in fact an A-W of syndromes, although she could have made it an A–Z by including Zellweger's syndrome! There are also a few minor problems, such as the absence of any discussion of renal biopsy in Alport's syndrome, the omission of epilepsy as a complication of neurofibromatosis, and a rather unclear distinction between Finnish nephrotic syndrome and the many other causes of nephrotic syndrome. There is also no reference to the finding of an expanded triplet repeat causing fragile X syndrome, explaining the unusual inheritance. The index contains a list of signs and symptoms found in different syndromes, similar to that seen in Gorlin's *Syndromes of the Head and Neck*, ostensibly as an aid to diagnosis. In fact, this volume is better used as a reference once a diagnosis has already been made. The lack of photographs also makes this volume more suited to the role of a lay reference work, rather than a diagnostic aid.

Overall, this is an excellent reference book for a wide range of health and educational professionals. It provides clear clinical information, and can give a quick snapshot of a condition for many people involved in the care of children with rare disorders.

ANDREW GREEN

Maternal Genetic Disease. Edited by N B Isada, A Drugan, M P Johnson, M I Evans. (Pp 272; £65.95.) Stamford, Connecticut: Appleton and Lange. 1994. ISBN 0-8385-6164-0.

People who advise pregnant women need to keep up with developments in genetics. Parents always want to know the risks of passing a condition to their children and whether anything can be done to reduce these. Often the first person they ask is their obstetrician or midwife. This book, edited by a distinguished team from the United States and Israel, aims to provide the information required. It has some good features but these are outweighed by many faults.

The book opens with six chapters on general aspects of genetic diseases, including specifically preconception counselling, chromosomal problems, and mental retardation. These deal with these problems in a similar way to most textbooks of genetics, albeit very briefly and with some important omissions. The book then changes character, and in the remaining 12 chapters a range of authors each tackle the genetic aspects of a specific maternal pregnancy problem, including the main medical problems that occur in pregnancy, renal, cardiac, haematological, neurological, and psychiatric disease, etc. This is a nice idea since, for generalists caring for pregnant women, these multifactorial conditions are much more common than the single gene defects on which most traditional genetic texts concentrate, and some chapters are very successful. However, for some diseases, once it has been stated that the inheritance is multifactorial, and the empirical recurrence risk given, there is little more to say. Unfortunately, this has not deterred contributors from padding out their chapters with platitudes, irrelevances, and repetitions, and the whole book cries out for stronger editing. The arrangement also leads to oddities. For example, cystic fibrosis (CF) appears only in chapters on anaesthesia and gastrointestinal disorders. The reason for the former appears to be that having written about malignant hyperpyrexia and succinylcholine sensitivity, the author needed a couple more pages to make a full chapter. The description of CF in the gastrointestinal chapter is concerned almost entirely with the important but rare problem of pregnancy in an affected woman. It is a quite inadequate guide to the day to day problems surrounding CF counselling and prenatal diagnosis for normal women with or without a family history.

There are many other omissions and imbalances. Some are serious and others simply rather strange. For example, myotonic dystrophy gets only five lines in one of the introductory chapters while multiple sclerosis gets 10 pages later on. It is unacceptable for a large genetics text in 1996 to omit any explanation of the whole area of triplet repeat sequences and genomic imprinting. Less serious, but still curious given the relative weight allocated to common multifactorial diseases, is the omission from a three page description of pre-eclampsia of any mention of the familial pattern of this disease. Readers will not learn that many experts even believe, albeit wrongly in my view, that this fascinating and common condition might be inherited in simple mendelian fashion, and that a number of groups are already doing gene linkage studies. They should be told.

This book bears all the hallmarks of being dashed off by busy authors and editors with more imporatant calls on their time. I cannot recommend it.

J G THORNTON

The Molecular Biology and Pathology of Elastic Tissues. Ciba Foundation Symposium 192. (Pp 361; £49.95.) London: Wiley. 1995. ISBN 0-471-95718-6.

This book contains the published proceedings of an excellent Ciba Symposium on the molecular biology and pathology of elastic tissues held in Kenya in 1994. As one expects of Ciba Symposium proceedings, the book is beautifully produced and very portable. Furthermore, it has been published within less than 12 months.

North American dominance in the field is very evident with 72% of the chapters and a

similar proportion of the participants originating from either the USA or Canada.

Here is an in depth coverage of the entire field of elastic tissue biology with state of the art information on topics such as biophysics, ultrastructure, elastin gene organisation, modifying enzymes, transgenic mice with elastin deletions, and the microfibrillar protein fibrillin 1. Coverage is also given to elastin regulation, histological changes in hypertension, and tumour biology. The range of topics mirrors most of those recently addressed in the 1995 Gordon Conference on the same topic. This book, of course, is available to a very wide audience whereas the Gordon Conferences, by their very nature, have a very limited audience.

Two important additions would have made this book even more valuable to an even wider audience. Firstly, a chapter on elastin related diseases would have been extremely valuable. While these are mentioned at various places throughout the book, the molecular pathology of disorders such as Williams syndrome, supravalvular aortic stenosis, cutis laxa, Marfan syndrome, PXE, Buschke-Ollendorf syndrome, aortic aneurysms, emphysema, etc, would have been a very valuable focus. After all, the first Williams mutations were described in 1993 and Marfan syndrome analyses have exploded since the gene sequence was published in 1991. Secondly, it would have been useful to have had a separate chapter on microfibrillar components other than lysyl oxidase and fibrillin. Microfibrils are important components of elastic fibres and include proteins such as MAGP, MFAP1, collagen VI, etc, in some of which the gene organisation resembles the fibrillin.

Nevertheless, the book will be of substantial interest to basic scientists interested in the field of connective tissue biology and also contains much interesting information for clinicians researching into connective tissue biology. Those with specific syndromic interests, however, are perhaps less well catered for, but the book is certainly a valuable addition to departmental libraries and large medical libraries should buy a copy.

F M POPE

Genetics and You. John F Jackson. (Pp 92; $9.50 pb.) USA: The Humana Press. 1995. ISBN 0-896-03330-9.

The aim of this short book is "to provide basic information about genetic disorders in a useful form for those who have no special training in genetics". Unfortunately, it fails to do this, giving genetic information in dry, technical terms and failing to address the wider, relevant issues. The starting point is medicine, rather than people's lives.

I will give some examples which speak for themselves. The chapter on prenatal diagnosis fails to address the difference between screening and diagnostic tests, the meaning of positive and negative results, or the psychological issues of anxiety and false reassurance. We are told: "The very slight risk of complications for mother and fetus from CVS is slightly higher than for amniocentesis. Technical error is higher for CVS". We are not told what these risks are nor what their implications are. But we are told: "There is probably a higher risk for Rh sensitisation by CVS, requiring use of Rho GAM injections for R-negative mothers". Only the term CVS is explained.

Carrier risk is explained: "The carrier risk for an apparently normal sibling of an affected person married to a nonrelative becomes 2/3, times the population carrier rate for the spouse, times one in four for an affected child if both parents are carriers. For CF, for example, the risk would be $2/3 \times 1/5 \times 1/4 = 1/150$, or in card cutting terms: one ace of hearts in three decks of cards."

The chapter on the new genetics starts with RFLPs and PCRs and goes on to explain why genetics is relevant to organ transplantation: "There is a series of genes lined up on chromosome 6 called the major histocompatibility complex (MHC) or simply the HLA region. There are separate genes at five loci, with an enormous number of possibilities. The HLA-A locus has at least 20 different genes, and the B locus has more than 40. There is an additional series of genes at the C, D, and DR loci." The issues of gene transfer and cloning are equally obscure: "Sometimes it is harmful, as in the transfer of a gene from animal to virus to make an oncogene capable of causing cancer. On the other hand, the entry of a single-cellular organism into the cells of higher organisms during evolution produced the mitochondrion", and "The cloning of a human analogous to that of frogs could be possible down the road."

Social and ethical issues are hardly referred to: "Heated debate has resulted from screening programs in the past and much has been learned". We are not told what the debate has been about nor what has been learned.

I make no apology for giving these quotes, the product of a clinical geneticist of 30 years' experience trying to communicate to the non-geneticist. While this review is unlikely to encourage people to buy this book, I hope it will encourage them to think about how we can better achieve the increasingly important task of communicating about genetic issues to non-geneticists. This is a multidisciplinary task, and would benefit from the expertise of those involved in the social and behavioural sciences, the public understanding of science, developing educational curricula, as well as communications using computer software.

SUSAN MICHIE

NOTICE

Fifth Annual Meeting of the International Genetic Epidemiology Society

This meeting will be held on 16–18 August 1996 in Rio de Janeiro, Brazil, in conjunction with the 9th International Congress of Human Genetics. For further information, please contact Dr Ruth Ottman, Columbia University, New York, telephone (212) 305–9188, fax (212) 305–2426.

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.

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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*, Box 238, Level 3, Laboratories Block, Addenbrooke's Hospital, Hills Road, Cambridge CB2 2QQ, UK 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 agree to their names appearing 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.

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Figures should be kept to a minimum and should be numbered consecutively in Arabic numerals. Legends should be typed on a separate sheet.

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