BOOK REVIEWS

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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 posed to the child. 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.)

Diagnosis of a rare condition in a child


People who advocate prenatal 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 intellectual 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 some very useful chapters on preconception, renal, cardiac, haematological, neurological, and psychiatric disease, etc. This is a useful 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, irrelevancies, 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 problems. The reason for this is hard to be that having written about malignant hypopryrexia and succinylcholine sensitivity, the author needed a couple more pages to make a full chapter. The description of CF might therefore be shorter than is necessary, and 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 triplet repeat sequences and genomic imprinting. Less serious, but still curious given the 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 a 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 daffed off by busy authors and editors with more important calls on their time. I cannot recommend it.

ANDREW GREEN


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

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


those medical interests, however, have been researching an important component of disorders such as Williams syndrome, PXE, VI, supravalvular aortic stenosis, cutis laxa, Marfan syndrome, and the 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 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. 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


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 x 1/5 x 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 clonning 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

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.