Some time ago, when a non-medical friend had a baby with Down's syndrome and asked me for a book to read, I was hard pressed to recommend one. Happily, this would no longer apply, as this excellent little book fits the bill admirably. The book is the latest in a series which aims to inform the lay public about common medical problems, and is intended primarily as a guide for parents of a child with Down's syndrome.

Attitudes to Down's syndrome have changed markedly in the last 20 years: more children are reared at home, and there are better opportunities for education and integration into the community. The growth of parents' associations, and the philosophy of normalisation, treating the disabled adult appropriately for his or her age rather than as a child, have both helped to bring about these changes. This book, written by a developmental paediatrician, is in tune with this philosophy but maintains a realistic view of the achievements of persons with Down's syndrome.

The book offers comprehensive advice on problems from infancy to adulthood. Parents describe their reactions and feelings at the time of diagnosis in their own words, and ways of coping at this difficult time with partners, other children, relatives, and friends are suggested. The features of Down's syndrome and its chromosomal basis are discussed. The wide range of developmental progress in Down's syndrome is illustrated, and ways of stimulating the child at different ages are outlined. The recommended guidelines for health maintenance, while not always routine in this country, for example, annual tests for hypothyroidism, argue for high standards of general health care. The complications of Down's syndrome, such as congenital heart disease, are well covered, as are behavioural problems and the techniques of behavioural modification.

The chapters of the book which deal with education and services available for children with Down's syndrome are of necessity rather general, as the book is sold in North America and Australia as well as the UK. Assessment, early intervention, and preschool groups are discussed, and parents are advised of the educational options available and of how to ensure that their child receives appropriate schooling.

The final section is concerned with adulthood, dealing with adolescence, sexuality, socialisation, employment, independence, and ageing. The situation in the UK with regard to the civil rights of intellectually disabled adults is compared unfavourably to other countries which have guardianship legislation. Controversial treatments, such as plastic surgery, sensory integration programmes, and diets are sensibly evaluated. The last chapter concerns the management of future pregnancies and the book ends with useful addresses and an index. Although there is no bibliography, other self-help books for parents are recommended in the text.

The book is well written, with clear explanations of technical terms, and is illustrated with diagrams, drawings, and attractive photographs. I would have no hesitation in recommending this book to parents. It should find a place in every special care baby nursery for the use of parents and staff, and in every child development centre, paediatric department, and genetic department.

Christine Garrett


This publication consists simply of a list of references to published articles in human cytogenetics and related disciplines. For the reviewer, at least, there is some merit in this approach, as it presents a rather simpler task than would be otherwise be the case!

The bibliography is arranged into 10 sections which include: a historical perspective; the chemistry and morphology of the chromosome; techniques; chromosome function; chromosomes in division; a general section on nomenclature, chromosomal variability, and abnormality; clinical cytogenetics; and chromosomes in evolution. The last section is a list of books and periodicals from which most of the references have been gleaned. The sections are extensively subdivided so that specific references may be located more easily. For example, the clinical cytogenetics section is subdivided into abnormalities of sex chromosomes, abnormalities of autosomes, spontaneous abortions, chromosomes and cancer, chromosome damaging agents, prenatal diagnosis, and genetic counselling. Most sections begin with more general publications to introduce the reader to the subject. In each subsection the references are arranged in alphabetical order.

I found the overall arrangement satisfactory and the bibliography is undoubtedly an easy way to get into the literature quickly and should be a useful aid for those new to the field and students. However, there does appear to be a preponderance of older references, possibly at the expense of more recent ones. The author admits that the list is selective, but the basis for omitting some good articles was not entirely clear to me. In addition, the assignment of some references to subsections seems a bit arbitrary at times. The inclusion of articles on dicentric X chromosomes and fragile X under chromosome polymorphism, together with similar errors involving autosomes, is perhaps indicative of lax editing or even incomplete knowledge of the subject. Some replication of references is also evident. Furthermore, the main value of a bibliography, in my experience, is to obtain ready access to recent publications. Indeed, the author recognises this, in that annual updates are planned. Nevertheless, the main body of the bibliography contains references only up to 1986; more recent papers up to 1988 are included in a large supplement, which curiously includes some much older references. This arrangement, which may well be because of factors beyond the control of the author, detracts somewhat from the practical value of the book.

Clearly this publication would be a valuable addition to the departmental library. As a means of information...

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retrieval, however, it has some fairly obvious limitations and it compares rather unfavourably both with computerised retrieval systems, some of which are far more flexible and efficient, and in some respects with specialised books that contain extensive bibliographies.

S H Roberts


This book is arranged in four sections. The first is a brief introduction to inheritance, and to the impact of molecular genetics on the field of neurological and neuromuscular disorders. The second section, more than one third of the book, describes the language and tools of molecular genetics. The third section, half the book, consists of chapters from distinguished research workers outlining their fields of work and describing their own recent contributions. The final section looks to the future, with discussions of genetic therapy and of the ethical issues thrown up by the advances in molecular genetics.

The second section is designed as a primer of molecular genetics. I found it to be a succinct outline of the field that would serve as a refresher to those whose acquaintance with genetics had slipped, but it would be difficult to use it as a primer for the completely uninitiated. The density of information is necessarily too great for it to be digestible as a primer, and some topics are touched upon only sketchily. This is inevitable, given the constraints of space, but it has prevented the editors from realising their stated aim. To cover all of genetics from the structure of DNA, through gene expression, DNA polymorphisms, library construction and analysis, and on to linkage analysis in just 170 pages is perhaps not possible. The quality of the contributions is generally high, and the editors have obviously taken great care to ensure that the techniques mentioned later in the volume are described here.

The third section is the heart of the book, and covers many inherited neurological disorders: for only some of these disorders has the corresponding gene product been identified. Each chapter describes the clinical disorder briefly, outlines the state of molecular knowledge, and then describes in more detail the work undertaken by the author(s). As a set of summaries of genetic knowledge (in most cases, as of early 1987) these are well written and will remain of interest for some time. What makes the volume exciting, however, is the description of the contributors' own work. In some chapters this is not emphasised, but I most enjoyed reading the accounts of work in progress or recently completed. The chapters describing genes whose product is known were generally longer and of more interest to me than were the others. The chapter on familial amyloidotic polyneuropathies, for example, was a lucid synthesis of molecular genetics, protein chemistry, and immunocytochemistry. The descriptions of some of the other disorders, those of unknown gene product, are perhaps less likely to retain their appeal. This is only to be expected, with the waning of the age of genetic linkage studies: a similar book in the future will have many fewer diseases in which the gene product remains unknown, or the gene locus uncertain.

I would warmly recommend this book to medical geneticists whose knowledge of scientific neurology warrants updating, and to neurologists and other clinicians interested in the genetic basis of neurological disorders. However, I would suggest some preliminary reading of elementary molecular genetics for those with little previous exposure. Medical students with an interest in the field, and hopefully with some background in molecular genetics, might also find some of the essays to be suitable general background reading.

Angus Clarke


This well known Directory was first published in 1968 to assist with medical communication in the field of birth defects. Specifically, it was envisaged that it would aid research by allowing identification of cognate researchers in other centres and also with the provision of counselling to distant branches of a family, particularly where they were resident in another country. The current edition contains information on 873 genetics units, but while this statistic is impressive, it serves to highlight marked deficiencies in genetic services in many countries. Of the 873 entries, 476 relate to centres in the USA and the remainder are distributed among 50 other countries with strikingly only four entries for the African continent and only a single entry for China. This imbalance might reflect a degree of underascertainment and this is in evidence for the UK where seven Regional Genetic Centres are not included, but I suspect the main problem is the apparent lack of genetic services in many countries throughout the world. Successive editions of this Directory will thus be required as this shortfall is remedied and I believe that we can all play a role in helping to make the Directory comprehensive by indicating omissions and duplications to the editors. It would also be helpful to have Fax numbers for the centres and I wonder if the option of supplying information on a computer disc would be worthwhile, particularly as the number of centres throughout the world expands.

The editors are thus to be commended for taking on this task which is of benefit to all of us who are involved in both research and service aspects of genetics. Copies of this new edition are available directly from the March of Dimes Birth Defects Foundation for a small fee which covers packaging and transportation.

J M Connor


The cover of this compact hard back volume describes it as "the first attempt to gather information on all aspects of Fanconi anemia in one source." The cover also describes the book as a monograph which suggests an unusual interpretation of this term in view of the inclusion of 57 contributors. The