Firstly, Nyhan reviews the classical approach to treating metabolic disorders by dietary restriction, replacement of a deficient product, or the employment of alternative pathways. There have been many successes in this approach, most notably with phenylketonuria. Secondly, Sweetman considers the treatment of certain metabolic disorders by various co-factors, perhaps the simplest and most straightforward approach. Unfortunately, disorders amenable to such treatment are all very rare. Thirdly, the use of chelation and other methods of toxin removal are discussed by Schneider. Here the best example is the use of penicillamine in Wilson's disease. Though highly effective in many cases, some patients prove intolerant and a new drug (triethylene tetramine dihydrochloride) may now be helpful and is currently under trial. Fourthly, the seemingly most obvious approach of enzyme replacement is reviewed by Hershfield. So far, however, there has been little if any success in this field for a variety of technical and biological reasons. Fifthly, organ transplantation for genetic disease is covered by Parkman. This approach has claimed moderate success with a number of disorders, most notably bone marrow transplantation in severe combined immunodeficiency disease (SCID). In many other disorders where this approach has been tried, including the mucopolysaccharidoses, the results are less clear. However, organ transplantation can occasionally provide interesting insights into pathogenesis. In cystinosis, for example, patients have been successfully treated with kidney transplantation though later there is histological recurrence in the grafted organ, showing that renal involvement is only a manifestation of a systemic disease.

Finally, the editor reviews gene therapy, an area in which he himself has been interested for many years, but this approach still remains only a possibility. "There are many technical issues still to sort out and to understand. What remains now is to solve those problems and to undertake the not inconsiderable task of making it work." Similar sentiments regarding other so called 'novel' approaches to treating genetic disease have been frequently expressed. We can only hope that the situation will change. Perhaps drug therapy may in some cases prove to be the best approach after all.

ALAN EMERY

Genetics and Human Nutrition. Eds Philip J Randle, John Bell, James Scott. London: John Libbey. 1990.

This volume is the fruit of an International Symposium organised by the Rank Prize Funds in March 1990. It covers a wide spectrum of topics ranging from diabetes to favism and many of the authors are distinguished experts in their fields. In their foreword, Professor D J Weatherall and Dr J Bell articulate the hope that some of the advanced genetic techniques which have helped unravel the mysteries of major monogenic diseases may soon be applied successfully to the more common metabolic disorders discussed in this book.

Several chapters are devoted to various aspects of diabetes and insulin resistance. One MHC encoded genetic element is known to determine approximately 20 to

30% of the genetic component of type 1 diabetes, but other genetic determinants have yet to be identified. Various approaches to identifying these loci are described. Once this is achieved the challenge will then be to understand the interaction between genes and environment which produces the susceptibility phenotype. Other topics covered include the protein kinase C pathway, protein serine/threonine phosphatases, fructose intolerance, islet prohormones, gut hormone and neuropeptide physiology, adipin, apolipoprotein B and lipoprotein (a).

This pot-pourri volume is well presented and clearly illustrated and provides a valuable overview of the genetics of some of the major metabolic disorders. Inevitably, some recent advances are not included but that is also a hopeful sign since it indicates that progress is being made in our understanding of these complex disorders. As a reference book for scientists and clinicians this volume would be a useful addition to a departmental library.

RUTH MORGAN

Jablonski's Dictionary of Syndromes and Eponymic Diseases. By Stanley Jablonski. (Pp 665.) Florida: Krieger. 1991.

The best part of this large volume is the brief introduction, which discusses the development, use, and misuse of the term 'syndrome' in lucid terms. Sadly, but predictably, the book itself does not live up to this. This compilation of diverse material will astonish even the clinical geneticist used to coping with heterogeneity, but much is irrelevant (for example, almost one page on 'headache syndrome' and 'malabsorption syndrome') while the entries on the true malformation syndromes are too brief and uncritical to be of use. Photographs are poor. The listings of those people describing syndromes are likewise too brief to be useful (for example, 'Young ID, British Physician'), while many entries ('hospital hobo', 'yuppie disease') are either obscure or bizarre.

I can recommend this book as light holiday bedside reading for the obsessional dysmorphologist (though I did not dare to send it to any one of them to review). On reflection, though, perhaps this collection is not so far removed from our own present attempts to classify dysmorphic syndromes, and should teach us not to regard them as too weighty or definitive.

PETER S HARPER

The Diffusion Of Four Prenatal Screening Tests Across Europe. Margaret Reid. (Pp 133.) London: The King's Fund Centre. 1991.

This is one of three books looking at factors involved in what is termed the 'diffusion' of three types of medical technology across Europe. The first two books concern themselves with renal lithotripsy and organ transplantation, the latter primarily considering liver and heart transplantation. This book, the third in the series, considers four prenatal

screening tests: amniocentesis, chorionic villus sampling, maternal serum α -fetoprotein sampling (for both neural tube defects and Down's syndrome), and ultrasonography.

The first chapter outlines the four different tests and their spread temporally and geographically within the European community countries. This is followed by four chapters with detailed summaries of reports from representatives of four of the individual member countries, the Federal Republic of Germany, Sweden, Greece, and Portugal. The four countries were felt to differ in geographical, social, economic, legal, and religious factors which might have affected any differences seen in the spread of these tests within those countries. The final chapter analyses the generic factors which were felt to affect the diffusion of the three types of medical technology.

It is suggested that an understanding of the factors such as the compatibility of the technology with ethical beliefs (for example, acceptability and legality of termination of pregnancy), the presence or absence of local or national policy decision making organisations (which might look at 'costs versus benefits'), the existence of a clinical infrastructure (for example, the establishment of clinical genetics services), and the 'observability' of a technique or procedure (for example, heart transplant versus prenatal diagnosis) might allow prospective determination of the appropriate allocation of medical resources and support. As acknowledged, however, the diffusion of new techniques or procedures often follows the technological imperative, "if it can be done, it should be done".

While this book is of interest in detailing the recent history of the innovation of these four types of prenatal screening tests in Europe, the analysis of the factors which might affect the subsequent diffusion seems relatively pedestrian, does not often determine which are primary or secondary, and, although a short book, it is, on occasion, repetitive in the material covered.

R F MUELLER

The New Genetics and Clinical Practice. 3rd edition. By DJ Weatherall. (£15.00.) Oxford: Oxford University Press. 1991.

When this book first appeared in 1982, it formed a modestly produced volume based on a lecture series, issued by a little known publisher. Despite this, its impact was immediate and profound, so that it is fair to say that the book and its author have been one of the principal influences in determining how molecular genetics has affected and been incorporated into clinical medicine. Nearly 10 years later, this third edition has grown and changed, but it still retains much of the original message and enthusiasm which were responsible for its success.

The book is now attractively produced and illustrated, remarkably inexpensive (reflecting its intended wide readership), and more comprehensive in its coverage of the field. Indeed, it now makes an excellent introductory textbook for students or clinicians, though its increased complexity may well deter some of those non-genetically orientated and educated clinicians that would have been influenced by the first edition.

This edition also shows how much wider the impact of molecular approaches has become; while haemoglobin disorders are still used prominently as examples, the advances in numerous mendelian and non-mendelian disorders are well covered and illustrate the convergence between medical genetics and the various specialities using molecular techniques. The potential and actual ethical problems are covered with both sensitivity and common sense; the applications to our understanding of cancers and the possibilities of gene therapy are fully explored.

Much of the original influence of the book was because the author was not only a scientist but a practising clinician, enabling him to act in a remarkable way as a bridge, not only for molecular concepts and techniques to become applied to clinical problems, but also for other clinicians to cross and to see for themselves the extraordinary possibilities for their own fields of work. The need for a bridge of this kind remains very great; one senses the author's frustration at what the past decade has not achieved, as well as what it has. In particular, the need for society as a whole to be scientifically and genetically literate has become imperative if the 'New Genetics', having firmly become part of clinical practice, is to be fully accepted by society for the ways in which it influences patients and families with many important and common disorders.

PETER S HARPER

London Neurogenetics Database. M Baraitser, R M Winter. (£395.00 plus optional annual updates. Available on 5.25" or 3.5" discs. For IBM microcomputers.) Oxford: Oxford University Press. 1991.

Those familiar with the *Dysmorphology database* built up by the same authors will understand both the aims and workings of the *Neurogenetics database*, which was compiled in order that clinicians involved with neurological disorders and syndromes would have ready access to reports of similar patients in medical publications. The authors also intended that the *Neurogenetics database* would be comprehensive and quick to use. These aims have been admirably achieved.

The Database is amazingly easy to install and it is easy to use AS LONG AS YOU READ THE MANUAL FIRST. This is small, compact, and clear. Some uses of keys

are unexpected, and some have different uses at different levels, so a little study beforehand is essential. The manual explains the different ways in which you can search for syndromes, or search for references if you so wish, or add in your own patient data. This latter feature is very useful for those of us who frequently see undiagnosable patients.

The *Database* is concerned with syndromes rather than common conditions, and complicated varieties of, for example, spinal muscular atrophy or spastic paraplegia are easier to find than the more common uncomplicated forms. Nearly 2000 syndromes with 8000 references are included, and many of these belong to just single patients. It is remarkably comprehensive to include such isolated cases, and saves the user much time and effort in searching through published reports. However, such conditions may in fact never be seen again in another patient.

Firstly, the user has to learn how to select a few conditions out of the 2000, and the art here is of choosing relatively uncommon features to lead into the syndrome search. Classified features are listed in the lucid and brief manual that accompanies the *Database*. For example, it is more useful to choose 'cystic changes' than 'dementia', although choosing both features on separate lines (which means add) is just as good. There are 229 syndromes which manifest dementia, and 100 with 'sparse hair'! Having found syndromes that you wish to learn about, you can ask for clinical features, abstract, and references. The abstracts (which are dated) are the high spots of the *Database*. They are composed by the authors and express their opinions as to the significance of the syndromes, how they relate to other conditions, and generally are wise and experienced assessments. For those disorders in which DNA techniques help carrier detection or prenatal diagnosis, the latest information is provided. If you wish to keep a record of these valuable comments and the appropriate references, you just have to request 'Print'; it does not matter what printer you have so long as it is connected to your computer.

The authors should be congratulated on what must surely be a very altruistic service, namely reading and assessing the body of neurological publications on behalf of the rest of us, and I do not envy them their "regular review of over 1000 journals". I am sure that many others, like me, will find it most rewarding to search through the *Database* for an unusual combination of signs, will thereby learn about many disorders other than the one being searched for, and will, in

passing, be familiarised with neurological publications. At the same time it is reassuring to know that the features of an undiagnosed patient are not already listed as a reported syndrome.

How often the *Database* is used will depend upon how often patients with rare syndromes are seen. Therefore, clinicians who deal with neurological syndromes will find the *Database* more useful than those who predominantly deal with common disorders, which is why paediatricians will find it more valuable than adult neurologists. Clinical geneticists will find it a helpful adjunct to the *Dysmorphology database*. One *Database* for each centre should be sufficient, particularly in view of the expense (£150) of the yearly updates.

SARAH BUNDEY

NOTICES

Ehlers-Danlos Support Group

The Ehlers-Danlos Support Group has produced an information booklet on Ehlers-Danlos syndrome. This has 20 pages and 15 sections covering various aspects of the syndrome. The authors of the booklet are Professor P Beighton, Professor A C Bird, Professor R Grahame, Mr A P Barabas, Dr H A Bird, Dr F M Pope, and Mr I P Hunter. The first copy of the booklet is free with subsequent copies costing £1.00 plus postage. They can be obtained from The Ehlers-Danlos Support Group (Mrs V A Burrows), 2 High Garth, Richmond, North Yorkshire DL10 4DG. Tel 0344 57695.

European School of Medical Genetics

The Fifth Course of the European School of Medical Genetics will be held on 5 to 12 December 1992 in Sestri Levante (Genoa), Italy. Directors: Victor A McKusick, Baltimore, and Giovanni Romeo, Genoa. Enquiries to: Istituto G Gaslini, Lab di Genetica Molecolare, L.go Gerolamo Gaslini, 5, 16148 Genova-Quarto, Italy. Tel: (010) 5636-370/400.