Firstly, Nyhan reviews the classical approach to treating metabolic disorders by dietary restriction, replacement of a deficient product, or the application of an alternate pathway. 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 tereamine 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 Pagean. 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 inconceivable 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 things will change. Perhaps drug therapy may in some cases prove to be the best approach after all.

ALAN EMERY


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 diet and disease to fadism 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 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 the current topic. 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 neuroendocrine physiology, adipin, apolipoprotein B and apolipoprotein (a).

This pot-pouri 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


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 uncutural 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 obsessive dysmorphologist (though I did not dare to send it to any of them for 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


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 for policy decision making. However, the discussion of 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 little value 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


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 this 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 on, 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 (reflecting its intended wide readership), and more comprehensive in its coverage of the field. Indeed, it now makes an excellent introduction to the subject for students, although its increased complexity may well deter some of those non-genetically orientated and educated clinicians that would have been influenced by the first edition.