

to Stephen Hawking's *A brief history of time*, which is also a best seller and which attempts to do for cosmology what Dawkins does for natural selection. For anyone with an interest in theoretical biology written in an interesting and seductive manner this is a must.

ALAN EMERY

Biochemical Genetics: A Laboratory Manual. Emmanuel Shapira, Miriam G Blitzer, James Miller, Diane K Africk. (Pp 145; £19.50.) New York, Oxford: Oxford University Press, 1989.

This book makes no pretence of being anything more than a bench book on biochemical genetics, as illustrated by its contents and general quality of presentation. Part I deals with enzyme assays, describing general procedures and buffers, and methods for specific enzymes using fluorimetric, spectrophotometric, and radioisotope techniques. It should be noted that all but two of the methods are for lysosomal enzymes, indicating a distinct bias in the contents of this section, which presumably reflects the specialised interests of the authors.

Part II is on diagnostic tests for metabolic disorders, specimen collection, and handling. There are sections on urine spot tests, thin layer chromatography of a number of types of metabolite, quantitative amino acid analysis, organic acid analysis by gas chromatography, and free and esterified carnitine assays. An appendix tabulates the clinical presentation of a range of disorders, including those of lysosomal storage, the urea cycle, and amino acid and organic acid metabolism.

There are obvious reservations about a book of this nature. One is that it is a simple recipe book of methods in use in the authors' laboratory and there is no critical appraisal of the techniques. Moreover, some of these methods, that for quantitative amino acids for example, are only valid for one particular type of analyser, and the information is of very limited value for a laboratory with alternative instrumentation. A second is that it might encourage a belief that methods like chromatography of amino acids and organic acids, and interpretation of the results, are straightforward matters which are learned easily. Recent

experience of quality control of urine amino acids in the UK has shown that this, and similar procedures, should not be undertaken lightly.

In conclusion, it is difficult to imagine that this book will have a wide appeal. Most established workers in this field would want to search widely before introducing a new technique. In general, it is accepted that biochemical genetics is best concentrated in centres where expertise already exists and newcomers, who might be attracted by this book, should be relatively few in number.

JOHN HOLTON

Human Gene Mapping 10. 10th International Workshop on Human Gene Mapping. (Pp 1147; £122.10.) Basel: Karger, 1989.

The First International Human Gene Mapping Workshop (HGM1) was held at Yale University in 1973 and the proceedings appeared in a slim monograph listing 31 confirmed autosomal assignments and offering a tentative map of 13 loci on the X chromosome. Sixteen years and a revolution in molecular biology later, the Tenth Workshop returned to Yale and has generated a tome to dwarf any telephone directory containing information on some 5000 mapped genes and DNA markers. This is essential reference material for the increasing numbers of scientists working on the human genome, as well as for those providing a service for patients with genetic disorders.

The information is presented in various ways to make it accessible to the reader. Committees assigned to individual chromosomes have produced reports summarising the data and highlighting additions and changes. The committee on clinical disorders and chromosome deletion syndromes has collated the information in a format which clinicians will find useful. Almost a third of the publication is taken up with the catalogues of 3400 DNA segments and 945 cloned genes. Abstracts of workshop presentations constitute another large section. One bar to communication and source of irritation is the exclusive use of D numbers throughout the text, necessitating repeated reference to the index of DNA segments. Since most

researchers use probe names in their everyday work, it would be helpful if these could also be given, perhaps in brackets.

Past workshops have managed to keep abreast of the increasing amounts of data on DNA markers and physical mapping, but presentation of data on genetic distances has been haphazard and incomplete. For a DNA marker of interest, one could readily find details of the probe, vector, and polymorphism, probably some indication of the cytogenetic localisation, and perhaps some ordering information, but seldom details of the genetic distance from nearby markers or disease loci. To meet this need, HGM10 includes summary scores from Bronya Keats's database of pairwise lod scores taken from published reports and workshop submissions. These scores, together with information on order derived from multipoint linkage maps and physical mapping data, have been used to construct genetic maps for each chromosome. This commitment to the compilation of linkage data is most welcome and would be all the more useful if the source of the primary data could also be listed.

HGM10 was made possible by an on-line database which allowed entry and modification of data by the 26 workshop committees and facilitated publication of the final report in only three months, a remarkable achievement. More sophisticated computerisation will be needed as the volume of information increases and there may well be changes in the way the data are contributed and validated by the scientific community. Certainly the format by which the compiled data are made available will have to change as the remaining 97% of the genome is mapped, otherwise we shall all need to reinforce our book shelves.

JOHN R. W. YATES

Genetic Variants and Strains of the Laboratory Mouse. 2nd ed. Ed Mary F Lyon, Anthony G Searle. (Pp 876; £90.00.) Oxford: Oxford University Press, 1989.

A brief period in the late sixties, studying as an undergraduate in Hans Gruneberg's department, taught me that mouse geneticists are different

Biochemical Genetics A Laboratory Manual

Frits A. Hommes



Biochemical Genetics A Laboratory Manual:

Biochemical Genetics Emmanuel Shapira, Miriam G. Blitzer, James B. Miller, 1989 The most comprehensive laboratory manual of its kind this book summarizes the common assays and laboratory procedures used for the diagnosis of genetic biochemical disorders Written in cookbook fashion it provides detailed descriptions of the procedures currently used in the diagnostic work up of patients with underlying inherited inborn errors of metabolism General procedures and buffer preparations are discussed Fluorometric and spectrophotometric assays for lysosomal and non lysosomal enzymes radioisotope methods and tests for metabolic disorders such as qualitative spot tests and thin layer chromatography are included For each assay the authors list the tissues to which it has been applied control ranges for leukocyte and fibroblast preparations and commercial sources of substrates Clear and concise this guidebook is designed to be accessible even to those with limited experience and will be an invaluable aid for genetics laboratories and medical geneticists **Techniques in Diagnostic Human Biochemical Genetics** Frits A. Hommes, 1991 Here is an up to date review of procedures currently in use in diagnostic biochemical genetics laboratories around the world Offers not only accounts of methodology but also provides guidelines for the interpretation of both standard and abnormal results The text includes coverage of most of the methods being employed to determine specific analyses as well as discussions of statistics and data management and the protocols of transmitting laboratory results with genetic information Many of the chapters contain introductory sections describing background information on the development of a particular genetic test and an evaluation of the clinical significance and applicability of the test **Practical Genetic Counseling for the Laboratory** McKinsey L. Goodenberger, Brittany C. Thomas, Teresa Kruisselbrink, 2017 This is an essential manual for the future of genetic counselling It codifies the theory and practice of laboratory genetic counselling in an accessible and comprehensive format With contributions from laboratorians geneticists and genetic counsellors from more than 30 institutions it offers a manual of standards and practices that will benefit students and counsellors at any career stage *Pediatric Reference Ranges* Steven J. Soldin, Carlo Brugnara, Edward C. Wong, 2003 The fourth edition of Pediatric Reference Ranges is a valuable reference providing instant and accurate reference ranges for chemistry and hematology analytes in an alphabetized user friendly format Reference ranges are provided for many new analytes such as dihydrotestosterone estrone iodide pregnenolone and zinc protoporphyrin Several new platforms have also been added such as Dade Behring RxL DPC IMMULITE and Sysmex **Laboratory Guide to the Methods in Biochemical Genetics** Nenad Blau, Marinus Duran, K. Michael Gibson, 2008-05-31 This manual deals specifically with laboratory approaches to diagnosing inborn errors of metabolism The key feature is that each chapter is sufficiently detailed so that any individual can adopt the described method into their own respective laboratory Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases Nenad Blau, 2003 Delay and mistakes in the diagnosis of inherited metabolic diseases may have devastating consequences Reference laboratory data are

scattered and clinical descriptions of rare conditions are hard to locate This book describes 298 disorders grouped into 35 chapters according to the type of condition Within each group of disorders chapters provide tables of pertinent clinical findings as well as reference and pathological values for crucial metabolites Relevant metabolic pathways and diagnostic flow charts are included There are four indices to make the book as user friendly as possible Disorders index Signs and symptoms index Organs index and Tests index The Physician s Guide provides paediatricians and other physicians with a unique aid to help them select the correct diagnosis from a bewildering array of complex clinical and laboratory data The book includes a CD ROM with search function *National Library of Medicine Current Catalog* National Library of Medicine (U.S.),

Current Catalog National Library of Medicine (U.S.),1993 First multi year cumulation covers six years 1965 70 **A Clinical Guide to Inherited Metabolic Diseases** Joe T. R. Clarke,2002-07-11 A clinically organized user friendly handbook on inherited metabolic diseases and their diagnosis **Potter's Pathology of the Fetus and Infant E-Book** Enid Gilbert-Barnes,Raj P. Kapur,Luc Laurier Oligny,Joseph R. Siebert,2007-05-01 This comprehensive reference addresses all aspects of fetal and neonatal pathology including complicated pregnancies multiple pregnancies abortion placental pathology and disorders affecting the full term neonate A consistent organization allows for quick access to specific guidance and nearly 2 500 illustrations 2 350 in full color depict conditions and abnormalities as they present in practice facilitating diagnosis An Image Bank on CD ROM new to this edition features all of the illustrations from the 2 volume set downloadable for presentations Offers comprehensive coverage of all common and rare embryonic fetal and infant disorders in one source Correlates clinical pathologic and genetic findings for each systemic disease Emphasizes the genetic and molecular basis of birth defects Features nearly 2 500 illustrations 2 350 in full color which depict each abnormality or condition as they present in practice Presents practical information on autopsy techniques and protocols Provides the latest guidance on molecular pathology immunohistochemistry DNA technology and more Offers an expanded discussion of developmental biology related to the pathogenesis of birth defects Features user friendly summary tables and diagnostic flow charts making information quick and easy to find Includes a CD ROM featuring all of the illustrations from the 2 volume set

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