Genetic Disease: Screening and Management

This multi-author text is based on the XVIth Birth Defects Symposium held in 1985 and as stated in the preface its purpose is to "help fill the information gap by focussing on many relevant and yet unanswered questions about genetic disease screening and management". To this end the book has chapters contributed by participants from the symposium outlining their personal experience and discussing various aspects of newborn screening for cystic fibrosis, sickle cell disease and other haemoglobin variants, metabolic disorders, hypothyroidism, and congenital adrenal hyperplasia as well as screening in pregnancy to identify haemoglobinopathy carriers. Particularly useful chapters are those by N Holtzman concerning the criteria and evaluation of screening programmes and that by H Levy reviewing the experience and management of maternal PKU.

One criticism of some chapters of the book is that they appear to be direct transcripts of talks as given at the symposium with little alteration when put into the written form, resulting in a feeling that some of the experience is anecdotal rather than distilled. The typeface used for the text and figures reinforces this feeling.

This book will obviously be of more interest to biochemical geneticists and to this end a number of the chapters go into considerable detail comparing different assay procedures with respect to their relative sensitivity. From the perspective of a clinical geneticist this is a book to peruse selected chapters of interest rather than to read from cover to cover.

R F Mueller

Monoclonals and DNA Probes in Diagnostic and Preventive Medicine

The publication of meeting proceedings with enticing titles such as Monoclonals and DNA Probes in Diagnostic and Preventive Medicine is an underhand attempt to help finance the meeting (BIOTECH RIA 86), which becomes a tax on libraries. This slim volume contains 24 very wide ranging contributions, most, but not all, of which discuss the use of DNA probes or monoclonal antibodies or both, although not necessarily in an overtly clinical context. In that sense, several chapters might be considered challenging to the reader who is prepared to sit and think about how the principles of the work described might be used in preventive and diagnostic medicine. The too familiar format of introductions, materials and methods, results, and discussion, to which most of the authors cling, is not conducive to imparting ideas and principles clearly. One of the few daring departures from the usual formula is a paper, appropriately from the Paris Pasteur Institute, on molecular genetic approaches to the problems of vaccine production. With enviable clarity, it presents in uncluttered terms the new possibilities being explored. Technical details are accessible from the two dozen references cited.

Several other contributions do make important, oft forgotten points. 'Molecular Approach to Diagnostic and Preventive Medicine: the Status of the Art' is the unexpectedly stilted title to an enjoyable opening paper by Siniscalco and colleagues who discuss clearly the utilisation of linkage in genetic analysis in families and populations, the problems of recombination and of genetic heterogeneity, and the advantages of looking at these problems at the ultimate DNA level. In several other cases (for example, use of oligonucleotide probes for detection of haemoglobinopathies) the illustrations are given without a clear statement of the principle. A more general approach would also be beneficial in fast moving fields such as muscular dystrophy where, inevitably, new data have succeeded those presented here.

On the monoclonal antibody front, it is difficult to patch together a coherent picture of the many currently feasible and potential uses for this prolific technology. Too much detail of experimental data obscures the exciting prospects that can be envisaged for anti-idiotypic antibodies. A couple of simple diagrams might help. Only Colnaghi et al address systematically the problems and possible solutions of tumour therapy with antibodies. The now mundane but numerically most frequent use of monoclonals in routine immunoassay is barely touched upon.

It is difficult to discern the type of audience for whom a book like this is aimed. I think that the majority of the papers leaves the general principles beyond the reach of well informed 'outsiders' such as clinicians. The cognoscenti might appreciate
some neatly made points, but will recognise them from regular ‘reading of the literature’. We are therefore left with the question of why the proceedings of such meetings need to be published. If money must be made by inveigling libraries to buy alluring titles, at least the participants should be encouraged to make their written contributions more general and more relevant to the title.

Veronica van Heyningen

The Balance of Improbabilities: A Scientific Life

There seem to be essentially two ways of writing about the history of science, either through discoveries and developments themselves or through the persons concerned with such events. The latter approach is often appealing because it adds a personal dimension and at the same time the reader may well be able to identify with many of the scientist’s problems and tribulations. This present volume falls into this latter class. It is an autobiography of Henry Harris whose work on cell biology is internationally renowned.

From relatively humble beginnings in New South Wales, through a series of scholarships, he entered Sydney University where he studied the Arts (modern languages and literature) and then medicine. He found the first part of the medical course frustrating because so much had to be committed to memory, but was stimulated by Fulton’s Physiology of the Nervous System, which I too found an exciting escape from much of the rote learning in the preclinical years. His dislike of memory learning stayed with him throughout his life, preferring a more logical approach which paved the way to his career in experimental science. He published his first paper in the Medical Journal of Australia while a medical student, and is now ‘heartily ashamed of it’!

The real turning point in his career came when he was appointed a research fellow under Florey in the Department of Pathology at Oxford. Here he first studied chemotactic responses in leucocytes but soon turned his attention to RNA turnover in cells, a subject which became a life long interest. His discovery that there was significant turnover of nuclear RNA was made some 15 years before the demonstration of heterogeneous nuclear RNA (from introns and exons).

His later work on cell fusion which grew out of his interest in nuclear RNA was to have important applications in many fields of cell biology, particularly in regard to the phenomenon of malignant transformation.

After a brief spell in 1959 at the National Cancer Institute in Bethesda, and then a very productive and happy period at the John Innes Institute, he returned to Oxford to take up the Chair of Pathology on Florey’s retirement, and in 1979 became Regius Professor of Medicine, a position he still holds.

His life long interest in English literature and his wide reading are reflected in his writing which is always engaging and elegant. This scholarly and instructive biography is a delight to read.

Alan E H Emery

Advances in Human Genetics

This 16th volume in the series of Advances in Human Genetics continues in the tradition of timely, scholarly, and comprehensive reviews of selected topics in human genetics. There are five reviews, three of which could stand alone as monographs and are worth the price of the book in themselves. First, ‘Genetics of Lactose Digestion in Humans’ by G Flatz begins by suggesting that lactose intolerance is a symptom complex of diverse aetiology and then describes the physiology of lactose digestion and its role in human nutrition. A distinction is made between high and low lactose digestion capacity (lactase persistence and lactase restriction, respectively) and their formal genetics are developed. Second, in ‘Chronic Granulomatous Disease’, J T Curnette and B M Babor used this condition to explore the role of oxidants in microbicidal activity and of oxygen radicals in inflammatory diseases. Normal neutrophil function is discussed at length as a background to understanding CGD. The genetic emphasis is on distinguishing X linked and autosomal recessive CGD, the relationship between CGD and the cell phenotype, and, finally, the cloning of the CGD gene by ‘reverse genetics’ with its implications for future research and treatment potential. Third, the real jewel of this volume is ‘Steroid Receptors and Their Disorders’ by L Pinsky and M Kauffman. This is the longest review and should be required reading for geneticists and endocrinologists. The ontogeny, physiology, and biochemistry of steroids, steroid receptors, and the complexes they form are elegantly developed as a background for examining discrete genetic lesions and variations in androgen resistance. The role that study of these conditions has played in elucidating normal processes is well developed.

The shortest review is ‘Perspectives in the Teaching of Human Genetics’ by R Davidson and B