

volume). Again random spores can be used to save the researcher from tetrad dissection. Perhaps an early step in these more difficult protocols should be "Find yourself a yeast geneticist" which would save a great deal of time and effort for the researcher. Fortunately this is not generally a problem as most yeast geneticists would be willing to help.

EDWARD J LOUIS

Autosomal Dominant Polycystic Kidney Disease. Editors A Sessa, F Conte, P Serbelloni, S Milani. (Pp 192; \$197.50.) Basel: Karger. 1995. ISBN 3-8055-6090-7.

Occasionally, the organisers of scientific meetings are lucky, and the date of the meeting coincides with the announcement of a huge step forwards in research in that field. The Vimerate seminar on Autosomal Dominant Polycystic Kidney Disease took place in June 1984, just after Peter Harris and co-workers identified the PKD1 gene on chromosome 16, nine years after the gene was mapped. While the greatest excitement was focused on the news about the gene, in fact the other papers presented, each of which forms a chapter of the book, are in many cases also "state of the art" reviews. Early chapters consider the physiology of the kidney and the pathogenesis of renal cysts. Cysts are fluid filled structures, and below 200 µm in diameter they communicate with the tubules from which they arise. Above 200 µm, they are non-communicating structures, and continue to expand because of net fluid accumulation within their lumens. Cyst expansion leads to progressive renal failure, and if expansion could be halted, the renal function could be preserved. Recent studies have detected mitogenic activity in cyst formation by renal epithelial cells *in vitro*, and may have an important role in accelerating the proliferation of renal cyst epithelium in PKD.

The chapter on liver disease by Torres will be particularly interesting for nephrologists, as the frequency of cysts in ADPKD increases with age from 20% in the third to 75% in the seventh decade of life, and liver cysts cause significant clinical problems in some patients. A few even require liver transplantation.

Pirson's review of ADPKD associated intracranial aneurysms (ICA) will be of considerable relevance to any clinicians who have been considering the difficult issue of whom to screen for ICA and when to operate. About 4-10% of ADPKD patients have an asymptomatic ICA, but in only 14% of them is the aneurysm >6 mm, and about half of these patients have a positive family history.

Pirson concludes that it is reasonable, therefore, to restrict screening to young patients with a family history of ICA, and to operate only if the ICA is >7 mm diameter.

Screening by magnetic resonance angiography every five years is recommended, unless there are symptoms which warrant immediate investigation.

Grantham's chapter on "Ethical Issues and Genetic Counselling" contains a check list of arguments for and against presymptomatic testing and figures for the gene penetrance using ultrasound scanning at 10 and 30 years. No distinction is made, however, between presymptomatic screening in adults and children, and it is suggested that CT scanning (or even MRI) may be offered to young patients with normal ultrasound scans. While clinicians will all agree that it is in the patients' best interests to detect treatable complications early (for example hypertension and urinary tract infections), this could be achieved by measuring the blood pressure and culturing the urine, without the need to look specifically for renal cysts. It seems that there are big differences between the United Kingdom and the USA in the extent to which presymptomatic screening is pursued in childhood.

Overall, this is a well written and clearly illustrated book which will be of considerable interest and practical use to both clinical geneticists and nephrologists, and both these groups will be keenly awaiting the next anticipated step forwards by the molecular biologists, that is, the development of mutation detection tests to provide definitive diagnostic/presymptomatic tests. It is very unfortunate, therefore, that it is likely to be some time before this becomes generally available, because the PKD gene lies largely within a genomic region which is duplicated elsewhere in the genome, making the search for mutations particularly difficult. It is likely that new methods of examination will be required if rapid progress is to be made towards solving these problems.

FRANCES FLINTER

Molecular Genetics of Human Inherited Disease. Editor Duncan J Shaw. (Pp 252; £32.50.) UK: John Wiley. 1995. ISBN 0-471-93459-3.

This book is one of a series aimed at bridging "the gap between pure research in the biomedical sciences and its practical application in clinical medicine". The series editors cite two objectives for books in the series: to promote (1) the understanding of the molecular basis of human physiology and disease, and (2) new techniques for diagnosis and treatment.

There is a brief introductory chapter by Duncan Shaw which outlines the techniques used by clinical and molecular geneticists in the rapid progress that has been made to identify genes responsible for many of the single gene disorders. The disorders are then

dealt with in great depth in six of the remaining eight chapters. These chapters almost constitute a history book that catalogues the localisation, cloning, and identification of disease causing mutations for Duchenne/Becker muscular dystrophies, cystic fibrosis, Huntington's disease, fragile X, myotonic dystrophy, and neurofibromatosis type 1. They also address what is known about the function of the genes, ranging from seven pages for the CFTR protein to one sentence for the Huntington's disease protein. Diagnostic techniques for each of the disorders are discussed. It would have been useful to have a more uniform structure to the chapters; however, on the whole they are well referenced, with suggestions for additional reading.

Although I am not a graduate student of medicine, the reader at whom the book is aimed, I enjoyed, particularly, two of the chapters. The Genetic and Molecular Analysis of the Human Y Chromosome is an excellent, clearly written chapter by Jean Weissenbach. It encompasses sex reversal in mouse models and in humans, the mapping and identification of candidate genes for TDF, spermatogenesis, and Turner's syndrome.

The final chapter, The Genetics of Psychiatric Disorders, concentrates on the application of genetics to disorders that do not have simple, mendelian patterns of inheritance. Apart from describing the clinical syndromes of schizophrenia, manic depression, and the dementias, the approaches that will be used to unravel the genetic components of many common disorders, such as cancer and heart disease, are clearly and concisely explained. DNA marker studies are described (complete with a simple explanation of lod scores), as are association studies and family, twin, and adoption studies. Inevitably, this chapter is already out of date on the molecular genetic aspects of Alzheimer's disease, predating the identification of the presenilin genes. However, this is a minor detraction from a chapter that addresses the next major challenge for geneticists, the identification of the genetic factors that contribute to multifactorial disorders.

Returning to the objectives of the series, perhaps the only one which receives scant mention is that of treatment for the genetically inherited disorders. The exception to this is the chapter on cystic fibrosis, which considers gene therapy as one possibility. There is much talk of collaboration throughout the book. Approaches to treatment for many of the genetically inherited disorders will require the collaboration of clinicians and scientists from a broad spectrum of disciplines, who will no doubt document their findings in another book.

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