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Cystic Fibrosis: Current Topics. Volume 2. Editors J A Dodge, D J H Brock, J H Widdicombe. (Pp 354; £60.00.) Chichester: J Wiley. 1994.

This useful second edition is once again presented in three sections: genetics, cell biology, and clinical, representing the areas of expertise of the editors. Each has collected contributions from a number of distinguished authors who have made personal contributions to their fields and each editor permits himself one chapter. The style is remarkably even for a multiauthor book and diagrams and illustrations are clear.

In the genetics section, there is a very useful chapter on mouse models by Julia Doren, including comparisons of replacement (knock out) and insertional models, admitting the possible leakiness of the latter technique by reversion to wild type by recombination. The suitability of mouse models is argued, despite different proportions of different cell types in the lungs of mice and man. Cutting writes interestingly on the splicing and alternative splicing of the CF gene and his chapter provides a fascinating insight into the basic relationship of genes, their mutations, and the final gene product. Among others alternative splicing of exon 9 is well described with the important insight that only small amounts of CFTR may be needed for normal function and that compound heterozygotes for $\Delta F508$ and R117H, some with lung disease and some with no features of CF except congenital bilateral absence of the vas deferens (CBAVD), depend on whether the splice acceptor is of the 5T, or more efficient 7T variety. Brock's chapter on heterozygote screening is well balanced, though obviously from the perspective of one who believes in

screening. Coutelle gives an update on gene treatment using different vectors and Romeo offers some modern physiological explanations for heterozygote advantages but admits that he poses more questions than answers.

In the cell biology section, Bradbury describes reconstitution of CFTR into planar bilayers as an alternate to patch clamp and discusses the advantages of the technique in allowing intracellular organelle function to be studied, while admitting present easy contamination of the system by protein impurities. Biwersi writes on the functions of CFTR other than simply as a plasma membrane chloride channel; he discusses critically a number of other roles. CFTR localisation in various tissues is discussed by Cohn. He assumes the central role of the submucosal gland in the pathogenesis of the lung disease, simply because it is the only lower respiratory tract cell to express CFTR strongly. Welsh writes an interesting chapter mainly on the R domain in regulating CFTR and how it influences the Cl⁻ channel.

In the clinical section, since gene treatment is dealt with in the genetics chapters it remains disappointing to note how little the genetic and cell biology discoveries have influenced clinical care. The clinical chapters are largely well written discussions on pathophysiology: the role of respiratory virus infections, energy requirements, evolution of pancreatic and liver disease, the role of cytokines in aggravating cell damage, and a chapter on heart and heart and lung transplant. Egan discusses an emerging role for double lung transplant, the improvement in operation survival, and the need for a better understanding of the immune mechanisms which lead to bronchiolitis obliterans.

Many of those who read *Journal of Medical Genetics* will find this a very useful book. Its price may prevent many individual people from owning a copy personally but genetics departments ought to have one on their shelves and it should be held by most medical libraries. The book provides much information to those working on specific aspects of CF while providing a very useful overall insight. A valuable chapter in the next edition would be an exploration of pharmacological regulation of the basic defect.

MAURICE SUPER

Molecular Genetic Medicine. Volume 4. Editor T Friedmann. (Pp 188; \$49.95.) San Diego: Academic Press. 1994.

This latest volume in the series is the best so far. All five articles give up to date reviews of

their subject, but with enough background material to ensure that they will not date too rapidly.

Two articles provide model examples of the successful melding of information from molecular genetics with conventional biochemical approaches. In "Superoxide dismutase, oxygen radical metabolism, and amyotrophic lateral sclerosis" (Patterson *et al*) we are reminded that although the brain makes up only 2% of body weight, it uses 18% of respired oxygen: no wonder that thinking can be so exhausting. There follows a detailed analysis of the chemistry of oxygen radicals and the role played by superoxide dismutase (SOD) in their disposal. The discovery of SOD1 mutations in familial motor neurone disease is surely one of the most exciting discoveries in molecular genetics, and this article illustrates how it opens up new vistas of biochemical analysis, as well as suggesting further candidate genes. "Protein import deficiencies in human peroxisomal disorders" (Wiemer and Subramani) is a similarly sophisticated treatment of its subject. If, like me, you have never been quite sure what a plasmalogen was, you will find the answer here. They are 1-alk-1'-enyl-2-acyl-*sn*-3-phosphoglycerolipids, the main end products of ether lipid biosynthesis, and may have a role in scavenging free radicals (a nice link with the previous article). They are one of the characteristic products of peroxisomes, a fact that can be exploited in screening for peroxisomal mutants.

Two articles tackle more basic biological problems. "Genomic imprinting in humans" (Driscoll) is a well structured and readable review of imprinting phenomena, and would serve as a good introduction to the subject. "Broad picture" answers to questions like why imprinting occurs, how it is controlled, and what is its chemical basis are, however, clearly still some way off. "Impact of gene targeting on medicine" (Thomas) is a succinct summary of a very topical subject, one that is keeping the latest generation of mouse geneticists in gainful employment all around the world.

Finally, "Molecular genetics of blindness" (Black and Craig) is a comprehensive review of the current state of play in the genetics of ocular disorders. There is no single gene disorder that manifests so much genetic heterogeneity as retinitis pigmentosa, and relatively few of the genes responsible have been identified as yet. Clearly the retina will provide another superb opportunity to integrate information from the genetic and cellular approaches, and the excellent section on the anatomy and biochemistry of the retina will help to put this further work into context.

A O M WILKIE