
After 50 years of confusion, the last six years have greatly increased our understanding of cystic fibrosis (CF). This disease, the most common autosomal recessive genetic disease in Caucasian populations, is eventually lethal. It causes recurrent lung infections, an inability to digest food properly owing to gradual blockage of pancreatic ducts preventing enzyme release and causing destruction of the pancreas, and over-salty sweat.

The frequency of the disease has made it a prime target for the attention of hungry teams of molecular geneticists. Since the discovery in 1985 of the first polymorphic markers showing linkage to the CF locus, the race, recently won by groups in Toronto and Michigan, for isolation of the CF gene itself was on. Simultaneously, our understanding of the basic defect in CF has been advancing methodically along the electrophysiological pathway. A discovery in 1983 that showed a chloride ion transport defect in the epithelium lining the sweat gland duct of CF patients has generated a cohort of electrophysiologists busy dissecting the ion transport systems located in the apical membranes of epithelia lining the organs affected in CF.

The diversity of the scientific specialties involved and the lack of a common language generated the need for a book such as this. It will also provide a scientific overview of the field for members of the medical profession. The book is a compilation of chapters written by specialists in the different areas of study, drawn together under the editorship of Peter Goodfellow. The choice of authors is an imaginative one.

The opening chapter summarises the clinical features of CF, though it has a rather tired approach. This is followed by a straightforward account of treatment of the disease. These two chapters form valuable reading matter for cell and molecular biologists and others who are trying to design meaningful experiments to establish how the CF gene works, and also for the many scientists thinking about the possibility of correcting the basic defect in CF in vivo by gene therapy. The rest of the book consists of two chapters covering the major research areas and one on clinical applications. The first of the research chapters is an engrossing exploration of what is known about the ion transport systems in CF. It succeeds in presenting a highly complex subject in a way that non-electrophysiologists will understand. An introduction to the basics of epithelial cell ion transport systems is followed by discussion of the specific epithelia that have been shown to exhibit abnormalities in CF, namely those in the sweat gland duct and the airways. Other epithelial cell systems that may also help in understanding the CF gene defect are mentioned. The word of caution sounded at the end of the chapter, reminding us that there is still a great deal that we don’t understand about how the CF gene defect really affects cellular ion transport systems, is timely.

Regrettably, the next section, on the molecular genetics of CF, does not live up to expectations. Admittedly this must have been an exceptionally difficult chapter to write. Not only has this field moved rapidly over the past four years, but until the CF gene itself was finally isolated it remained difficult to assess which molecular approaches to the problem would merit recording for posterity. Having said this, the chapter starts well with an accurate and clearly written account of the recent history of CF molecular genetics. It does, however, assume that the reader has a certain knowledge of recombinant DNA technology. Classical and reverse genetics approaches to CF are explored, together with sections on pulsed field gel electrophoresis, jumping and linking libraries, and chromosome mediated gene transfer. However, the overall emphasis and approach is rather parochial. Furthermore, one of the key strategies in the successful cloning of the CF gene, that of screening cDNA libraries made from tissues expressing the basic CF defect (sweat gland duct epithelial cells), is not covered at all.

The final aspect of CF to be included in this book is prenatal diagnosis of the disease. Though well written, this chapter suffers greatly from the time lag between the writing and publication for a book of this sort. The importance of different prenatal diagnostic tests for CF has changed rapidly over the last three years. DNA based tests, combining linked markers and haplotypes for the markers pXV2c and KM19, which are in strong linkage disequilibrium with the CF gene, account for the vast majority of CF prenatal diagnostic tests. Microvillar enzyme assays now make up only a small percentage of these tests. The addition of an addendum has partly remedied this, but it results in undesirable fragmentation of the text. This chapter would have been enriched by speculation on the nature of population screening for CF by direct methods once the gene and its mutations had been defined.

This book will be valuable in parts to provide the background for current researchers into how mutations in the CF gene cause the pathological effects. However, the timing of its launch is more than a little unlucky, as the recent cloning of the CF gene has rendered much of the book seriously out of date.

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