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Book Reviews

Cystic Fibrosis: Horizons. Proceedings of the 9th International Congress, Brighton, England, June 9th–15th 1984. Edited by D. LAWSON. Chichester: John Wiley. 1984. 446 pages. £15.00. ISBN 0 471 90439 2.

Cystic Fibrosis (CF) is the most frequent severe autosomal recessive disease among Caucasians: 1 in 2000 children born has cystic fibrosis. The nature of the basic defect, the gene coding for the defective protein, or, failing that, a closely linked polymorphic DNA probe are hotly pursued problems on both sides of the Atlantic. Thus the work discussed at the latest major meeting on this disease should be of interest to many scientists and clinicians.

I must declare at the outset that for me this volume of papers and abstracts read like the book of the film as I had been a minor participant at the congress – a wallflower wilting by one of the 250 posters presented.

Following a deadline for camera-ready contributions only six months before publication at the time of the meeting, the book should represent the most up-to-date work in the field of CF research. However, as is frequently the case with reports of large international meetings, some of the invited contributions (and thus the longer papers in this book) are included for historical or political reasons rather than on current scientific interest. The organization into sections is somewhat arbitrary and the reader will need to make his own connexions between items on related subjects under different headings. The contents of the three sections – prenatal diagnosis and carrier detection, pathophysiology, and the basic defect – overlap in several areas. The last two sections are separated by the only distinct area: aspects of clinical management.

The first paper, by David Brock on prenatal diagnosis, describes a new test to be applied to pregnancies at risk. The data given are for retrospective samples of amniotic fluids collected in an earlier (failed) trial of a different system. Non-transferability of assay systems from one laboratory to another has been a major problem in CF research. This new technique was being tried out prospectively in the originating and several other laboratories at the time of abstract writing (p. 14 and posters 1.20, 1.22 and 1.23), but as human gestation cannot be squeezed into much under 9 months, the reader is left uninformed about the outcome of the pregnancies being followed. These results are clearly critical to evaluation of the new test but will have to emerge through normal channels in the journals.

While benchside workers are slaving over the validation of a suitable prenatal diagnostic test, an enterprising group of medical geneticists conducted a survey, among the parents of affected children in California, on the market for prenatal diagnosis capable of identifying CF foetuses. Michael Kaback's report shows a great desire for a reliable test which could, in conjunction with elective abortion, avoid the birth of further affected offspring. Without such a test more than 60% of couples with one affected child decided to have no further children. There is no indication whether these fascinating data are published elsewhere in more detail. I also ask myself how representative are the opinions of highly educated Southern Californian parents in this delicate area.

Much of the work on the pathophysiology of cystic fibrosis, which is primarily a disease of the exocrine organs, has been centred around exciting studies on ion permeability changes which have been observed by several workers in a variety of cell types studied. In many of these studies little consideration is evident of the genetics of autosomal recessive diseases. Very few physiological studies, two of which are in the 'basic defect' section, have attempted to study heterozygotes who may be expected to have intermediate

levels of normal or mutant gene product. Some statements elsewhere show a horrifying lack of understanding of gene expression: 'It [CF] is transmitted in what appears to be an autosomal recessive mode of inheritance (ref). Because of the latter, it would be expected that the basic defect would express itself in all the cells of affected individuals.'

The molecular biological approaches to tracking down the CF gene are optimistically included in the 'basic defect' section. Most of this work is at a very preliminary stage. Bob Williamson's admittedly contagious enthusiasm for the molecular solution to genetic diseases is based on success stories where the gene defect was well characterized before the cloners were let loose on the problem, as in the thalassaemias. Linked probes perhaps capable of tracking *X*-linked Duchenne muscular dystrophy or autosomal dominant Huntington's chorea are not yet in regular use for prenatal diagnosis and in neither case has the mutant gene been pinpointed. The CF gene has not even been assigned to a chromosome, so looking for linkage by exclusion using chromosome-assigned DNA probes with restriction fragment length polymorphisms is an uphill job in a recessive disease with no reliable heterozygote detection. Partly because genetics was not the primary concern of this congress, there was no discussion of the problem of whether even closely linked (1–3 cM) probes would be reliable enough for prenatal diagnosis. The enormity of the task of walking 10^6 to 3×10^6 base pairs to find and identify the actual CF gene is so far in the future with this approach that it does not frighten the intrepid molecular biologist.

More than half the book consists of up to one-page poster abstracts. These vary from tantalizing through pedestrian to incomprehensible summaries of what we all know involves many man-years of painstaking labour in most cases. It is, however, impossible to assess data from these vignettes except occasionally by digging out quoted publications – not an easy task when references of the style 'Smith A. B. *et al.*, submitted' are allowed.

Precious library allocations should not go on conference reports of this type. The most zealous workers in the CF field may feel the need to widen their 'Horizons' by borrowing a copy from a colleague or at worst from inter-library loan.

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Genetics and Development. By JAMES H. SANG. Harlow: Longman. 398 pages. £13.95. ISBN 0 582 44681 3.

Many years ago, while I was studying the inheritance of quantitative characters in Drosophila, this organism seemed to me to present most unsuitable material for an attack on that fundamental biological problem, the genetic control of development: not only did Drosophila consist of a succession of two very different organisms, but the transformation from larva to imago took place rapidly inside a magic box, the pupal case, which allowed very limited experimental access to its contents. This opinion has happily turned out to be a very superficial one, largely because of the stubborn attitudes of far-sighted geneticists such as E. B. Lewis, who burrowed away at the impenetrable mysteries raised by homoeotic mutants such as bithorax and Antennapedia, or sought out mutations which might relate the segmental patterns of larva and imago. These studies have provided excellent material for the new techniques of molecular genetics, and have now placed Drosophila in the forefront of the attack on the genetic control of development.

Sang has written a textbook of developmental genetics for advanced undergraduate and post-graduate students of biology, but he has achieved much more than this by writing a critical survey of the subject which will be of great value to everyone interested in what is both the major outstanding general biological problem and also a research area of intense activity and rapid progress. The fact that something like two papers a day relevant to this field are being published makes his book all the more useful, since he