BOOK REVIEWS


The Royal College of Physicians (London) report on Prenatal Diagnosis and Genetic Screening (RCP 1989, 'the screening report') represents a watershed in the development of genetic services in Britain. Traditional patient and family based medicine is confronted by the need for a community wide service for the prevention of genetic disease and radical solutions are required for a potentially vast expansion of screening and prenatal diagnosis.

The screening report noted that 2 to 3% of all couples are at high and recurrent risk of having a child with an inherited disorder. The burden of genetic disease is further illustrated by recent estimates from the Department of Health in British Columbia which suggest that 5-5% of the population will develop genetic disease by the age of 25, while 60% may do so in a lifetime.

To cope with the needs of genetic patients and families a network of regional genetic centres has been developed in Britain. These centres combine 'under one roof' clinical and laboratory diagnosis, counselling, and genetic registers for the investigation and support of the families. This arrangement follows the pattern of multidisciplinary integration advocated by the Joint Statement from the medical royal colleges in 1986, which was supported by the Department of Health in its reply and in subsequent ministerial statements.

However, new molecular genetic technologies will soon offer tests for all major genetic disorders and permit widespread carrier detection and prenatal diagnosis. Pilot studies for cystic fibrosis population screening are being initiated which, if successful, may eventually involve the entire population of reproductive age in Britain. The demand for genetic diagnosis, counselling, and screening can then be expected to rise rapidly, although this demand already exceeds the capacity of the current establishment of genetic centres. This is shown by another study by the Royal College of Physicians' which found the equivalent of 156-3 full time clinical genetic staff (including 37-58 consultants) in the UK, representing only 2-75 total clinical staff per million population.

The screening report placed particular stress on the importance of 'community genetics', a new term implying the exposure of persons at low prior genetic risk to population screening programmes. Community genetics is discussed in detail elsewhere in this issue of the journal by Dr Bernadette Modell, who was the Honorary Secretary to the working party that produced the screening report.

The recommendations of the screening report are wide ranging and have important implications for the future of patient and family based medical genetics. While clinical geneticists will appreciate the recommendation that: 

"... genetic screening and prenatal diagnosis services should be equally available to the whole community..." clinical geneticists may wonder if two of the recommendations taken out of context might inhibit specialised pre-pregnancy genetic workup and counselling, frequently necessary before prenatal diagnosis, and might create a new cadre of community geneticists remote from genetic centres. The relevant recommendations are:

"... (genetic screening and prenatal diagnosis services) should be recognised as an intrinsic component of maternal and child health services..."

"... specialist counsellors should be attached to each obstetric unit practising prenatal diagnosis...".

Clinical geneticists are concerned that few doctors have had appropriate undergraduate genetic education and may not yet have acquired adequate postgraduate training. This is shown most clearly by another Royal College of Physicians (London) report which indicated very variable teaching of genetics with a mean of 20 hours preclinical (range 2 to 66 hours) and 5-5 hours timetabled clinical teaching. Teaching was given by many different departments and was generally of uneven quality or absent.

The screening report recognised these educational deficiencies and recommended that:

"... professional training in medical genetics and the principles of genetic
counselling should be provided for all maternal and child health workers (GPs, obstetricians, paediatricians, family planners, health visitors, and midwives). Official contact should be made with the relevant professional bodies to develop the genetic component of the training curriculum and to organise updating courses for existing practitioners . . .

"... It is urgent to define a career structure for . . . specialist counselors, who may have differing professional backgrounds, and carry out a wide range of activities . . ."

To this should be added the recommendation of the Royal College of Physicians (London) genetic teaching report that:

"... implementation (of medical undergraduate genetics teaching) should be co-ordinated nationally by a Genetic Education Task Group to accelerate the pace of genetic education . . ."

The screening report hopes to ensure good standards of genetic care by recommending that:

"... though monitoring should be organised on a regional basis, a national centre is needed to develop appropriate methods, co-ordinate information nationally, and stimulate equal service delivery throughout the country . . ."

and by establishing that:

"... Face-to-face counselling and written information are complementary . . . one should not be given without the other.

The screening report is timely, has been well received, and will be influential, so clinical geneticists must give its recommendations full attention. Quite simply there is too much work for consultant clinical geneticists and their teams. Some of this new work may be more appropriate for others to do, but there is much to be said for joint planning by clinical geneticists and public health doctors.

Clinical geneticists have a central role in promoting genetic education and in constantly emphasising the right of individual persons to make their own informed decisions about parenthood. This principle could be forgotten in the enthusiasm for population screening.

RODNEY HARRIS


Following the advertisement in Nature, sometime last July, announcing the forthcoming publication of a volume on PCR technology I immediately placed an order at the bookshop. Then I waited. And waited. Last December it arrived. Was it worth the wait? Certainly. At last, almost everything you probably wanted to know about PCR in one volume.

The text is divided into three sections: basic methodology, research applications, and medical applications. Each of the 19 chapters, written by an expert in his, or her, particular field, is clear, concise, and well presented. A word of warning for the novice contemplating attempting PCR for the first time; despite giving an excellent introduction to the subject, this is not a laboratory manual. This is a comprehensive review of PCR, its applications and modifications, and is aimed, I feel, at those already familiar with the technique.

The first section gives a thorough outline of the principle of the technique, and the factors to bear in mind when designing an experiment for oneself. The properties of the enzyme Taq polymerase are described, as is the automation of the process which the isolation and cloning of this enzyme has permitted. Sample preparation is also covered and four rapid and simple protocols for different starting materials are given. Hopefully, the uninitiated student will have heeded the warning about contamination given in the introduction to this section. As these first four chapters are likely to be the most widely read, particularly by those embarking on setting up gene amplification, perhaps more emphasis should have been placed on means of avoiding contamination?

The second section is, basically, one of ideas and describes ways of manipulating the reaction or the reaction products. Thus, one is not limited to 'straightforward' amplification of a known sequence but can use the PCR product for cloning, sequencing, and identification of mutations, or can manipulate the primers, the known templates flanking the DNA, or RNA, of interest, to introduce new, or create altered, sequences. The wide variety of topics covered in this section certainly demonstrates the versatility of the technique. Disappointingly, methods of amplifying sequences when only a limited amount of information is available, for example, starting with only one specific primer or with protein sequence information, were not included. Most of the chapters in this section were accompanied by protocols and, where absent, suitable references cited.

The final section covers medical applications. The chapters on prenatal diagnosis, diagnosis of new mutations at known loci, and HLA class II gene polymorphisms were excellent. Have a look at figure 6 in chapter 15. Superb! However, the forensic applications of PCR were puzzling. After describing how one aims to achieve the unequivocal identification of a criminal suspect, and describing a number of suitable approaches, the case described failed to achieve a definitive result. The chapters on oncogenes and viruses were extremely interesting, particularly as both these topics are going to become so important in the future.

To summarise, this book is well written and well presented, and bearing in mind the difficulty of bringing together so much relevant material in such a rapidly evolving science, succeeds as the first major textbook of its type. I am sure this book will become a standard text on the bookshelves of many researchers in this field.

JENNIFER LYNCH