

Book Reviews

DNA Fingerprinting: State of the Science. Edited by S. D. J. PENA, R. CHAKRABORTY, J. T. EPPLIN and A. J. JEFFREYS. Basel: Birkhauser. 1993. 480 pages. Hardcover sFr. 178. DM 198. \$140. ISBN 3 7643 2781 2. Softcover sFr 118. DM 136. \$93. ISBN 3 7643 2906 8.

The 46 articles in this book were 'selected and prepared' from the 2nd International Conference on DNA Fingerprinting (Belo Horizonte, Brazil, November 1992), and are very broadly grouped under the three main headings 'Genomic organisation, dynamics and variability of tandemly repeated DNA sequences' (11 papers), 'Applications to the study of human populations' (15 papers), and 'Applications to micro-organisms, plants and animals' (20) papers.

Research progress in this field is very rapid and anyone with some background knowledge of DNA fingerprinting technology will find this book of absorbing interest. Not only are the methods of identifying individuals becoming steadily more sensitive, but new applications to new species are continually being applied, and in addition the nature of satellites, minisatellites and microsatellites, in fact of tandemly repeated DNA sequences in general in eukaryotic genomes, is coming under increasingly close scrutiny.

One aspect of natural interest is the progress of forensic methods applied to identifying rapists and rapist/murderers – of continued interest because problems in getting DNA fingerprint conclusions accepted in American and British criminal law cases still arise (see, for example, *Nature*, 13 January 1994, which discusses arguments between the FBI and US geneticists as to whether the US National Academy of Sciences should revise its very influential report on the use of DNA technology in forensic science, which is barely two years old. The FBI says 'Yes', the geneticists say 'No'. The same page of *Nature* discusses a case in London in which the Court of Appeal ordered a retrial of a rape case because one of the forensic scientists and the Judge had fallen into the so-called 'prosecutor's fallacy' – confusion between two methods used to interpret the significance of an apparent match between two DNA samples).

There is a limited amount of information in the book under review to help the reader decide on the

current forensic problems. Budowle & Monson, of the FBI Forensic Science Research Center, on pages 177–191 compare databases for 4 variable-number-of-tandem-repeat (VNTR) loci from 24 populations of different reference groups, such as American Blacks, Caucasians, southeastern Hispanics, etc., using fragment lengths obtained by Hae digestion. Their analysis suggests that errors from using a larger and more mixed database to estimate the chance that a suspect could have the same DNA profile as someone else in the community (who could therefore be the guilty one), would not be biased by using the 'wrong' database. Balazs (pages 194–210) analyses the DNA profiles of 14 ethnic groups using several VNTR loci and finds 'a high degree of similarity even among very different populations and that regardless of the population studied these loci are a powerful and very discriminating tool for human identification'. The populations in fact include Black, Caucasian and Hispanic from different areas, Chinese, Tobas, Maya, Pima, Cheyenne, Navajo and Australian Aborigine.

Jeffreys *et al.* (pages 125–139) are developing an alternative approach to DNA typing 'which combines the extreme variability of large minisatellites with the speed and sensitivity of PCR, and further to eliminate DNA fragment measurement from the procedure'. Their method (labelled MVR-PCR, where MVR stands for minisatellite variant repeats) performs MVR mapping on the minisatellite MS32 in the D1S8 locus. This is clearly a very powerful new technique which the authors consider to be 'sufficiently simple, sensitive and robust to be of great use in forensic analysis'. This is amply demonstrated by their paper, and my only caveat is that it may be difficult to get advocates, judge and jury to understand it. The sensitivity of MVR-PCR is indicated by unpublished results of the authors, who were able to type an individual successfully from saliva traces recovered from the back of a postage stamp. This is confirmed in a later paper by Holland *et al.* on 'Short tandem repeat loci: application to forensic and human remains identification' (pages 267–274), who quote Hochmeister *et al.* (1991 *J Leg med* **104**, 229) and Walsh *et al.* (1992 *J For Sci JFSCA* **37**, 387) as having identified DNA from a stamp licked by an embezzler and also from a cigarette butt left at the scene of the crime by a thief.

Numerous other papers are of great, even absorbing, interest and I can only refer to a few. Ginther *et al.* (pages 211–219, a paper with a very long title) examine mitochondrial DNA sequence variation and allele frequencies of several nuclear genes in Mapuche Indians from Patagonia and compare their genetic profile with other Amerinds and with worldwide populations, as part of the attempt to obtain data on the genetic characteristics of genetically unique populations which are in danger of disappearing. The next paper, by Roewer *et al.*, studies the Yanomami Indians of the Amazon lowland, who stayed in relative isolation until recent times and who have received anthropological field study.

The third main section of the book, which I have not yet touched on, discusses applications of DNA fingerprinting to plants (one paper), to micro-organisms, parasites and invertebrates (six papers), to fishes and birds (five papers) and to mammals (eight papers). These contain many interesting studies which deserve the reader's attention, but I will only mention one: Ruth & Fain's article on 'The "individualization" of large North American mammals' (pages 429–436), which discusses the application of DNA profiling to wildlife crime. Animal species in the world pet trade include at least 140 mammal species, 85 bird species and 57 reptile and amphibian species. The global market for illegal wildlife products has escalated into an estimated \$1–2 billion a year. Fish and wildlife estimates are \$200 million per year in the US alone, where it is estimated that two animals are poached for every animal taken legally. DNA fingerprinting has been applied to more than 150 forensic cases in North America, including such species as elk, deer, wolf, parrots, bighorn sheep, eagle and bear. It has also been used in more than 150 suspected fraud cases of horse paternity.

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Preconception and Preimplantation Diagnosis of Human Genetic Disease. Edited by R. G. EDWARDS. Cambridge University Press. 1993. 340 pages. Price £60/US \$99.95 hardback. ISBN 0 521 40425 8.

When the prospects for preimplantation diagnosis were first reviewed eight years ago by McLaren (1985) the outlook was pessimistic. Almost immediately afterwards, however, there was a surge of interest and research on preimplantation diagnosis began in earnest. The beginnings of the subject may be traced further back and include the early attempts to separate X- and Y-bearing spermatozoa, to control the sex of the conceptus, and the sexing of preimplantation rabbit embryos from biopsied cells (Gardner &

Edwards, 1968). Nevertheless, it was not until the late 1980s, when human *in vitro* fertilization had become securely established as a viable clinical technique, that research focused on the possibility of diagnosing genetic diseases in the human preimplantation embryo. Current research in preimplantation diagnosis stems from the previous achievements in reproductive biology, embryology and genetics. The mammalian embryologists, who had experience of handling preimplantation embryos, were the first to get involved and they shaped much of the early research.

The initial phase of research in preimplantation diagnosis involved feasibility studies. Mouse and human preimplantation embryos were used to compare different biopsy procedures, to evaluate different genetic tests etc. Sensitive biochemical assays were tested early in this phase but these have largely been abandoned in favour of DNA-based tests. *In situ* hybridization was tried first but this was superseded by PCR (polymerase chain reaction). PCR remains the method of choice for most studies but *in situ* hybridization seems to be making something of a come-back as FISH (fluorescence *in situ* hybridization). Undoubtedly there have been some indifferent experiments but, nonetheless, real progress has been made during this 'feasibility phase'. The next phase (clinical trials) began in 1990 at the Hammersmith Hospital (Handyside *et al.* 1990) but, as yet, few other centres have followed suit. Preimplantation diagnosis is a demanding business and it is not yet clear whether it will develop into a routine clinical service. We may now be at a watershed between the feasibility studies and clinical application. Some appropriate biopsy and analytical techniques have been identified but further refinements are needed before they can be used routinely.

So much for the history, now what of the book? Professor Edwards has earned a unique place in the field, through his pioneering work on *in vitro* fertilization. His enthusiasm for the further development of preimplantation diagnosis shows through in this volume: as well as editing the book he co-authored 4 of the 14 chapters. Despite the title, much of the book is devoted to providing background information about prenatal diagnosis in general. I found these review chapters useful but this information is readily available elsewhere. I suspect that this emphasis may disappoint those who have developed an interest in preimplantation diagnosis from a background of prenatal diagnosis rather than embryology or reproductive biology.

I must confess that I found the organization of the book a bit disconcerting. The 14 chapters are grouped into 5 sections entitled respectively 'Background' (chs. 1–3), 'Development of prenatal diagnosis' (chs. 4–6), 'Methods of DNA diagnosis' (chs. 7 & 8), 'Preconception and preimplantation diagnosis' (chs. 9–13) and 'Ethics and Law' (ch. 14). These sections