BOOK REVIEWS

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Preimplantation Genetics, edited by Yuri Verlinsky and Anver Kuliev, is the publication of the proceedings of the First International Symposium on Preimplantation Genetics held in Chicago in September 1990. The main emphasis of the meeting was preimplantation diagnosis, a promising new approach to the prevention of the birth of a baby with a severe genetic disease. Previously, the options open for a couple at high risk of having a diseased baby were to avoid having children or to face the possibility of abortion if prenatal diagnosis during pregnancy indicated an affected fetus. Preimplantation diagnosis provides the option of genetically testing embryos outside the body and initiating pregnancy by replacing in the woman’s uterus only embryos shown to be free of disease.

Preimplantation diagnosis requires safe and efficient methods of obtaining embryos by in vitro fertilisation (or possibly uterine lavage), biopsy of a single cell without compromising the viability of the embryo, genetic analysis of the single cell (for chromosome defects, gene mutation, or enzyme deficiencies), and replacement of the embryo in the woman’s uterus. It requires the cooperation and collaboration of experts: the clinical geneticist, the obstetrician, the embryologist, and the scientist. The state of the art in September 1990 is amply covered by this book, with its comprehensive coverage of these areas as well as the ethical and legal issues. At the end of the book there is the bonus of half a dozen technique papers. The manuscripts are printed directly for more rapid publication and it is a pity that the format is inconsistent and the presentation and clarity of data are variable in quality. Some papers are reviews, some appear to be direct transcripts of the talks, and some seem rather irrelevant to the main themes. Several papers appear not to have been edited. Throughout the book I noticed inconsistency in naming the preimplantation embryo as ‘embryo’ or ‘pre-embryo’. The term ‘pre-embryo’ was introduced during the debate on embryo research in Britain and, owing to this unfortunate timing, was seen by some as an attempt to ‘belie’ the status of the embryo. I note that ‘embryo’ is the designation now used by the Human Fertilisation and Embryological Authority in Britain.

This book serves as an introduction to many of the cast of characters in this field although not all the contributions at the meeting are included. The most recent and sophisticated single cell gene amplification is expertly covered in the book by Arneheim et al, who achieve high efficiency detection and identification of three loci in single sperm. Other papers I enjoyed were the exciting paper by First on the potential of embryo manipulation techniques as performed in farm animals, the comprehensive paper by van Steirteghem et al on clinical aspects of IVF and preimplantation diagnosis, the paper by Bank on somatic gene therapy, the thoughtful paper by Gordon on germline therapy, and the three papers on ethical and legal issues. Also, thankfully, Edwards provides an excellent and most necessary introduction describing early development and providing a good brief review of the field.

It is good to see papers on basic and applied research intermingling, showing clearly that the introduction of preimplantation diagnosis is far from being derived from the basic research in the science laboratories. However, we are left with a crucial question – does it work? In the excitement of the new sophisticated reproductive and genetic diagnostic technologies, we risk forgetting the main players, the couples at risk of having a genetically diseased baby who are contemplating this route for prevention. It is tempting for them to think that the ‘latest is the best’ and for us to think that because it is possible to do this that it should be done. Often, as in this book, the excitement is about the potential of the new technology in advance of its proven safety and efficacy. Unfortunately, preimplantation diagnosis so far in the period following September 1990 has not inspired us with confidence. In Britain, one in seven ‘sexed female’ embryos was an error resulting in a male fetus at risk of X-linked disease; in the US, one in five embryos diagnosed ‘free from disease’ and replaced to initiate pregnancy resulted in a fetus homozygous for cystic fibrosis, a disastrous result considering that one in four of the embryos of this couple would be expected to be normal or heterozygous (phenotypically normal) by chance alone. These early mistakes might suggest that the appropriate attention was not paid to quality control or that unforeseen error risks have not yet shown themselves. At this stage, the requirement for complex IVF procedures, the risk of diagnostic errors, and the low efficiency of pregnancy after embryo transfer might make preimplantation diagnosis less attractive than conventional forms of early prenatal diagnosis. Nevertheless, preimplantation diagnosis may take its place in the future and overall I recommend this book as a good introduction to an exciting new era in medical genetics.

Marilyn Monk


Medical Genetics in Canada traces the development of human and medical genetics in Canada from the beginning of this century to the present time. The first few chapters of the book deal with short biographies of four of the pioneers of genetics in Canada. This section is followed by historical accounts of the regional development of medical genetics in the various provinces, each chapter written by geneticists from the relevant geographical area. This section is of least interest to the general reader. What is of greater interest to a more general reading audience are chapters on the evolution in Canada of medical genetics as a specialty, and one on future challenges for medical geneticists in Canada. The first of these chapters deals with the struggle, over a 15 year period, for medical genetics to be recognised as an independent medical specialty. This experience has been shared by geneticists in other countries as well. The final chapter deals with problems confronting medical genetics communities in other countries, such as assuring adequate funding for the delivery of genetic services, and concerns regarding the commercialisation of human genetics research. Excepting these final chapters of the volume, this book would appear to have a limited audience, mainly consisting of medical historians and Canadian geneticists.

Jonathan Zonana


The 5th International Congress on early fetal diagnosis was held in Prague in July 1990. Not only did it attract a distinguished group of contributors from Western countries, but it was the first opportunity for Eastern European scientists, many of whom have financial difficulties in travelling to international gatherings outside their borders, to present their findings. This book, a substantial paperback of over 600 pages, records the proceedings of the Congress.

Virtually every topic encountered in modern prenatal diagnosis was covered at the meeting. There are sections on the epidemiology of congenital malformations, periconceptional prevention, in-vitro fertilization, genetic counselling and ethical problems, in addition to the hard core subjects of ultrasound diagnosis, cytogenetics, biochemistry, and molecular genetics. Professor Hans Galjaard, who has been heard to complain that he is becoming the obligatory closing speaker, this time opened the meeting with a masterly overview of the state of the art.

Conference proceedings are not one of my favourite forms of reading, since in most cases it is impossible to remember what was written in print. This one is rather different, simply because of the wealth of detail contained in its pages. It is attractively presented, well organised, and firmly edited. Almost the only flaw is the lack of a subject index; quite unnecessary, I would recommend it to any library interested in maintaining a comprehensive stock of books in this important and rapidly developing subject.

D J H Brock