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#### **BOOK REVIEW**



## Molecular Cytogenetics Protocols and Applications

Methods in Molecular Biology Volume 204. Editor Yao-Shan Fan. (Pp 424; \$135.00.) Totowa, NJ: Humana Press. 2002. ISBN 1-58829-006-9.

This book is a very up to date manual covering the background, methodologies, and applications of molecular cytogenetics techniques. The emphasis is on the diagnostic applications of FISH in the many areas of medicine on which it impinges, including paediatrics, fetal and reproductive medicine, pathology, haematology, oncology, and, of course, medical genetics. With 27 chapters and over 60 authors, all of whom are experts in their field, this book clearly shows how far molecular cytogenetics techniques have developed over the last two decades.

The book is divided into three parts. Part 1 covers the basic concepts and techniques. The opening chapter by the editor Yao-Shan Fan provides a very helpful overview of the scope of the book and an extensive set of references for further reading. The following chapters cover probe labelling (DNA and RNA probes) and basic FISH techniques. The second part of the book is devoted to evolving techniques and applications and includes chapters on microdissection, PRINS, SKY FISH, M FISH, CGH, colour banding FISH, fibre FISH, multitelomere FISH, fluorescence genotyping for telomeric regions, and microarray CGH. Special applications of molecular cytogenetic techniques in chromosomal disorders are covered in part 3 of the book. These include chapters on the application of FISH to the delineation of marker chromosomes and the diagnosis of microdeletion syndromes. Other chapters cover FISH interphase nuclei screening for prenatal diagnosis including preimplantation diagnosis and fetal cells in the maternal circulation, in addition to the interphase FISH screening of routine amniotic fluid samples. This section concludes with a chapter on the application of FISH and CGH in reproductive pathology. The fourth and final section of the book covers the application of molecular cytogenetic techniques to cancer diagnosis. Chapters include the use of CGH in cancer investigations and the application of interphase FISH for the BCR/ABL rearrangements in CML and for HER2 amplification in breast cancer. Also included in this section are the interesting combined approaches, firstly of chromogenic in situ hybridisation with FISH in pathology and secondly simultaneous fluorescence immunophenotyping and FISH on tumour cells.

With any multiauthor book, there are bound to be differences in approach to the writing of individual contributions. This book has taken a surprisingly consistent approach, perhaps an illustration of good editorial control. All of the chapters have good introductory sections and are well referenced, as well as containing the authors' preferred methodologies. Each chapter also includes a comprehensive notes section (effectively tips and troubleshooting advice from the experts). However, a major problem with the book is the lack of comparison between different molecular techniques. In the preface, it is suggested that the book should help the technologist or cytogeneticist determine which procedure to perform for an informative result. I am not sure that the book provides this, as most if not all of the contributors are bound to select their own preferred methodology. For example, in the chapter on FISH screening for telomere abnormalities, an otherwise excellent chapter by Samantha Knight and Jonathan Flint, the practical application of only one of the two commercially available probe sets for this test is covered. More importantly, elsewhere in the book there is a first class chapter on interphase FISH for prenatal diagnosis of common aneuploidies by Baruch Feldman et al. This covers the topic extremely comprehensively and provides 89 literature citations. However,

there is no mention, as far as I could see, throughout the book of the other alternative molecular approach of quantitative fluorescence PCR for prenatal aneuploidy detection. While it may be reasonable to excuse the editor by saying that the book is not designed to cover purely molecular genetic techniques, in the same section as the prenatal FISH, there is a chapter on the molecular detection of uniparental disomy. This chapter sits rather incongruously among the others, but is in itself a useful and important topic. Another chapter which seems to have lost its place describes the BAC resource for molecular cytogenetics. This is the final chapter in the book, which appears to have been added as an afterthought. Surely this should have been in evolving techniques and applications rather than "special applications in oncology". The oncology applications would also have benefited from more chapters, for example, haematological disorders other than CML, solid tumour FISH (other than HER2), and perhaps a chapter on the screening of urine samples for bladder cancer.

Perhaps these topics are covered in the companion volume (Methods in molecular biology, volume 220. Cancer cytogenetics: methods and protocols). Another criticism is that the provision of colour plates is very variable. Some chapters are well illustrated, others less so. For example, the chapter on SKY FISH relies on black and white illustrations, whereas the MFISH chapter has glorious full colour images. Furthermore, the overall size of the book is relatively small (16 cm by 24 cm) and the size of typesetting and tight layout does not make for easy reading when compared with, for example, Rooney's "Human cytogenetics: a practical approach".

On the plus side, there is an incredibly large amount of information packed into this volume, none of it superfluous. Although other textbooks that cover FISH techniques are available, this book provides a more comprehensive, up to date, and thorough coverage of diagnostic molecular cytogenetics than any of the other books currently available.

In summary, I would recommend it as a reference source for everyone working in and interested in the exciting field of diagnostic and research molecular cytogenetics.

**Lionel Willatt** 

150 **PostScript** 

### **NOTICE**

#### **European Human Genetics** Conference 3 - 6 May 2003 ICC, Birmingham, England

#### Plenary sessions

- Low Penetrance Genes and Cancer Susceptibility
- Public Issues Population DNA Banks
- · Recent Developments in Neurogenetics

#### Symposia

- Bioinformatics
- Stem cells
- Sensory genetics
- Alternative splicing
- Cancer genetics
- Alzheimer disease
- SNPs and haplotypes
- Chromosomes in genetic disease
- · Genetics and endocrine problems

#### Workshops

- Syndrome identification
- Cytogenetics
- · Problems in counselling/ethics
- Genotyping and mutation detection arrays— practical problems Quality control
- Prenatal cytogenetics
- · Community genetics

#### Abstract deadline

Will be via world wide web. Closing date 13 January 2003.

Further information available from: The Vienna Medical Academy of Postgraduate Medical Education and Research, Alserstrasse 4, A-1090 Vienna, Austria. Tel +43 1 405 13 83 22 Email: eshg@medacad.org

#### **CORRECTIONS**

In the October 2002 issue of the journal, in the paper by Van Maldergem et al (J Med Genet 2002;39:722-33), we regret that some of the authors' names and affiliations were inadvertently omitted. They were:

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In the July 2002 issue of the journal, in the Online mutation report by Olivieri et al (p e39), there was a missprint in table 2. For No 12 the mutation should have been 1031G>T instead of 1006G>T.

In the December 2002 issue of the journal, in the paper by Khoo et al (pp 906-912), all c.1732delTCinsAC mutations should read C.1732delTCinsA. This error occurs on page 906 (Abstract) and page 910 (Discussion).